NOVOCLINIC

Pharmacogenomics Report

John Doe (4376)

Patient Information

Patient name: John Doe
Patient ID: 4376
Date of Birth: 1990-05-02
Sample ID: CN/M/2024/05/293
Sex: Male
Contact: johndoe@email.com

Report Information

Date created: 04-06-2024

Summary of your results

Patient has one gene (CFTR) that has significantly altered function. Usage of Ivacaftor is not recommended. Alternate drug is required.

Review of specified medications

- Standard precautions: medication can be prescribed according to standard dosing guidelines with standard monitoring of medication effects
- Use with caution: medication can be prescibed with dose adjustment and/or with increased monitoring.
- Consider alternatives: medication should not be prescribed due to potentially reduced efficacy or increased toxicity.

Drugs	Implication	Recommendation
ivacaftor	CFTR: An individual diagnosed with cystic fibrosis (CF) and negative for a CFTR variant listed in the FDA-approved drug label as being responsive to ivacaftor.	Ivacaftor is not recommended
acenocoumarol	An INR ≥ 6, resulting in an increased risk of bleeding, occurs in 8-12% of these patients during the first weeks of treatment with standard regulation by the Anticoagulation Clinic. The genetic variation increases the sensitivity to acenocoumarol.	Monitoring by the ANTICOAGULATION CLINIC (National INR Monitoring Service): • recommend to use 50% of the standard initial dose OTHERWISE: • recommend to use 50% of the standard initial dose • recommend more frequent monitoring of the INR
azathioprine	Increased risk of thiopurine-related leukopenia, neutropenia, myelosuppression,Lower concentrations of TGN metabolites, higher MeTIMP, this is the 'normal' pattern. Normal risk of thiopurine-related leukopenia, myelosuppression.	Start with reduced starting doses (30%-80% of normal dose) if normal starting dose is 2-3 mg/kg/day (e.g., 0.6 – 2.4 mg/kg/day), and adjust doses of azathioprine based on degree of myelosuppression and disease-specific guidelines. Allow 2-4 weeks to reach steady-state after each dose adjustment (PMID 20354201, 11302950, 15606506, 16530532). **Other Considerations** Normal starting doses vary by race/ethnicity and treatment regimens. If standard dose is below normal recommended dose, dose reduction might not be recommended for intermediate metabolizers.
dexlansoprazole	CYP2C19: Normal PPI metabolism; may be at increased risk of therapeutic failure compared to CYP2C19 IMs and PMs	Initiate standard starting daily dose. Consider increasing dose by 50-100% for the treatment of H. pylori infection and erosive esophagitis. Daily dose may be given in divided doses. Monitor for efficacy.

lansoprazole	CYP2C19: Normal PPI metabolism; may be at increased risk of therapeutic failure compared to CYP2C19 IMs and PMs	Initiate standard starting daily dose. Consider increasing dose by 50-100% for the treatment of H. pylori infection and erosive esophagitis. Daily dose may be given in divided doses. Monitor for efficacy.
mercaptopurine	Increased risk of thiopurine-related leukopenia, neutropenia, myelosuppression,Lower concentrations of TGN metabolites, higher MeTIMP, this is the 'normal' pattern. Normal risk of thiopurine-related leukopenia, neutropenia, myelosuppression.	Start with reduced starting doses (30%-80% of normal dose) if normal starting dose is ≥75 mg/m2/day or ≥ 1.5 mg/kg/day (e.g., start at 22.5-60 mg/m2/day or 0.45-1.2 mg/kg/day) and adjust doses of mercaptopurine based on degree of myelosuppression and disease-specific guidelines. Allow 2-4 weeks to reach steady-state after each dose adjustment. If myelosuppression occurs, and depending on other therapy, emphasis should be on reducing mercaptopurine over other agents (PMID 20354201, 18685564, 8857546, 18987654, 20010622, 16401827, 11302950, 16530532, 9634537). If normal starting dose is already < 75 mg/m2/day or < 1.5 mg/kg/day, dose reduction may not be recommended. **Other Considerations** Normal starting doses vary by race/ethnicity and treatment regimens. If standard dose is below normal recommended dose, dose reduction might not be recommended for intermediate metabolizers.
omeprazole	CYP2C19: Normal PPI metabolism; may be at increased risk of therapeutic failure compared to CYP2C19 IMs and PMs	Initiate standard starting daily dose. Consider increasing dose by 50-100% for the treatment of H. pylori infection and erosive esophagitis. Daily dose may be given in divided doses. Monitor for efficacy.
pantoprazole	CYP2C19: Normal PPI metabolism; may be at increased risk of therapeutic failure compared to CYP2C19 IMs and PMs	Initiate standard starting daily dose. Consider increasing dose by 50-100% for the treatment of H. pylori infection and erosive esophagitis. Daily dose may be given in divided doses. Monitor for efficacy.
phenprocoumon	An INR ≥ 6, resulting in an increased risk of bleeding, occurs in 17% of these patients with standard regulation by the Anticoagulation Clinic. The genetic variation increases the sensitivity to phenprocoumon.	Monitoring by a ANTICOAGULATION CLINIC: • recommend to use 50% of the standard initial dose NO monitoring by a anticoagulation clinic: • recommend to use 50% of the standard initial dose • recommend more frequent monitoring of the INR

thioguanine	Increased risk of thiopurine-related leukopenia, neutropenia, myelosuppression,Lower concentrations of TGN metabolites, but note that TGN after thioguanine are 5-10X higher than TGN after mercaptopurine or azathioprine. Normal risk of thiopurine-related leukopenia, neutropenia, myelosuppression.	Start with reduced doses (50% to 80% of normal dose) if normal starting dose is ≥ 40-60 mg/m2/day (e.g., 20-48 mg/m2/day) and adjust doses of thioguanine based on degree of myelosuppression and disease-specific guidelines. Allow 2-4 weeks to reach steady-state after each dose adjustment. If myelosuppression occurs, and depending on other therapy, emphasis should be on reducing thioguanine over other agents (PMID 20354201, 11037857). **Other Considerations** Normal starting doses vary by race/ethnicity and treatment regimens. If standard dose is below normal recommended dose, dose reduction might not be recommended for intermediate metabolizers.
warfarin	The genetic variation results in increased sensitivity to warfarin. This results in an increase in the risk of excessively severe inhibition of blood clotting (INR > 4) during the first month of the treatment.	Use 60% of the standard initial dose. The genotype-specific initial dose and maintenance dose can be calculated using an algorithm, as used in EU-PACT: see https://www.knmp.nl/patientenzorg/medicatiebewaking/farmacogenetica. From day 6 on the standard algorithm without genotype information can be used to calculate the dose.
atazanavir	UGT1A1: Somewhat decreased UGT1A1 activity; low likelihood of bilirubin-related discontinuation of atazanavir.	There is no need to avoid prescribing of atazanavir based on UGT1A1 genetic test result. Inform the patient that some patients stop atazanavir because of jaundice (yellow eyes and skin), but that this patient's genotype makes this unlikely (less than about a 1 in 20 chance of stopping atazanavir because of jaundice). **Other Considerations** All studies correlating UGT1A1 genotypes with atazanavir adverse events have involved ritonavir boosting. However, concentration-time profiles are equivalent when boosted with either cobicistat or ritonavir (PMID 23532097), and bilirubin-related adverse events including discontinuation of atazanavir occur in a similar percentage of patients prescribed atazanavir with cobicistat or ritonavir (PMID 23532097). Associations between UGT1A1 genotype, bilirubin elevations, and atazanavir/r discontinuation therefore almost certainly translate to atazanavir/cobicistat.
desflurane	These results do not eliminate the chance that this patient is susceptible to Malignant Hyperthermia. The genetic cause of about half of all MH survivors, with MH susceptibility confirmed by contracture test, remains unknown (PMID: 28902675).	Clinical findings, family history, further genetic testing and other laboratory data should guide use of halogenated volatile anesthetics or depolarizing muscle relaxants.

desflurane	These results do not eliminate the chance that this patient is susceptible to Malignant Hyperthermia. The genetic cause of about half of all MH survivors, with MH susceptibility confirmed by contracture test, remains unknown (PMID: 28902675). A negative or inconclusive genetic test cannot be assumed to indicate normal RYR1-related phenotype and should be interpreted in context of clinical findings, family history and other laboratory data.	Clinical findings, family history, further genetic testing and other laboratory data should guide use of halogenated volatile anesthetics or depolarizing muscle relaxants.
enflurane	These results do not eliminate the chance that this patient is susceptible to Malignant Hyperthermia. The genetic cause of about half of all MH survivors, with MH susceptibility confirmed by contracture test, remains unknown (PMID: 28902675).	Clinical findings, family history, further genetic testing and other laboratory data should guide use of halogenated volatile anesthetics or depolarizing muscle relaxants.
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halothane	These results do not eliminate the chance that this patient is susceptible to Malignant Hyperthermia. The genetic cause of about half of all MH survivors, with MH susceptibility confirmed by contracture test, remains unknown (PMID: 28902675).	Clinical findings, family history, further genetic testing and other laboratory data should guide use of halogenated volatile anesthetics or depolarizing muscle relaxants.
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isoflurane	These results do not eliminate the chance that this patient is susceptible to Malignant Hyperthermia. The genetic cause of about half of all MH survivors, with MH susceptibility confirmed by contracture test, remains unknown (PMID: 28902675).	Clinical findings, family history, further genetic testing and other laboratory data should guide use of halogenated volatile anesthetics or depolarizing muscle relaxants.
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methoxyflurane	These results do not eliminate the chance that this patient is susceptible to Malignant Hyperthermia. The genetic cause of about half of all MH survivors, with MH susceptibility confirmed by contracture test, remains unknown (PMID: 28902675).	Clinical findings, family history, further genetic testing and other laboratory data should guide use of halogenated volatile anesthetics or depolarizing muscle relaxants.
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sevoflurane	These results do not eliminate the chance that this patient is susceptible to Malignant Hyperthermia. The genetic cause of about half of all MH survivors, with MH susceptibility confirmed by contracture test, remains unknown (PMID: 28902675).	Clinical findings, family history, further genetic testing and other laboratory data should guide use of halogenated volatile anesthetics or depolarizing muscle relaxants.
sevoflurane	These results do not eliminate the chance that this patient is susceptible to Malignant Hyperthermia. The genetic cause of about half of all MH survivors, with MH susceptibility confirmed by contracture test, remains unknown (PMID: 28902675). A negative or inconclusive genetic test cannot be assumed to indicate normal RYR1-related phenotype and should be interpreted in context of clinical findings, family history and other laboratory data.	Clinical findings, family history, further genetic testing and other laboratory data should guide use of halogenated volatile anesthetics or depolarizing muscle relaxants.
succinylcholine	These results do not eliminate the chance that this patient is susceptible to Malignant Hyperthermia. The genetic cause of about half of all MH survivors, with MH susceptibility confirmed by contracture test, remains unknown (PMID: 28902675).	Clinical findings, family history, further genetic testing and other laboratory data should guide use of halogenated volatile anesthetics or depolarizing muscle relaxants.
succinylcholine	These results do not eliminate the chance that this patient is susceptible to Malignant Hyperthermia. The genetic cause of about half of all MH survivors, with MH susceptibility confirmed by contracture test, remains unknown (PMID: 28902675). A negative or inconclusive genetic test cannot be assumed to indicate normal RYR1-related phenotype and should be interpreted in context of clinical findings, family history and other laboratory data.	Clinical findings, family history, further genetic testing and other laboratory data should guide use of halogenated volatile anesthetics or depolarizing muscle relaxants.
azathioprine	The guideline does not provide a description of the impact of a normal metabolizer phenotype on azathioprine.	The guideline does not provide a recommendation for azathioprine in normal metabolizers.
lansoprazole	The guideline does not provide a description of the impact of a normal metabolizer phenotype on lansoprazole.	The guideline does not provide a recommendation for lansoprazole in normal metabolizers.

mercaptopurine	The guideline does not provide a description of the impact of a normal metabolizer phenotype on mercaptopurine.	The guideline does not provide a recommendation for mercaptopurine in normal metabolizers.
omeprazole	The guideline does not provide a description of the impact of a normal metabolizer phenotype on omeprazole.	The guideline does not provide a recommendation for omeprazole in normal metabolizers.
pantoprazole	The guideline does not provide a description of the impact of a normal metabolizer phenotype on pantoprazole.	The guideline does not provide a recommendation for pantoprazole in normal metabolizers.
thioguanine	The guideline does not provide a description of the impact of a normal metabolizer phenotype on thioguanine.	The guideline does not provide a recommendation for thioguanine in normal metabolizers.
warfarin	N/A	N/A
warfarin	The guideline does not provide a description of the impact of a normal metabolizer phenotype on warfarin.	The guideline does not provide a recommendation for warfarin in normal metabolizers.
allopurinol	The guideline does not provide a description of the impact of the ABCG2 rs2231142 GG genotype (c.421CC; p.141QQ) on allopurinol.	The guideline does not provide a recommendation for allopurinol in patients with the the ABCG2 rs2231142 GG genotype (c.421CC; p.141QQ)
amitriptyline	Normal metabolism of tertiary amines,n/a	No recommendation
atomoxetine	CYP2D6: n/a	No recommendation
atomoxetine	CYP2D6: n/a	No recommendation
atorvastatin	SLCO1B1: Typical myopathy risk and statin exposure	Prescribe desired starting dose and adjust doses based on disease-specific guidelines. **Other Considerations** The potential for drug-drug interactions and dose limits based on renal and hepatic function and ancestry should be evaluated prior to initiating a statin.
capecitabine	DPYD: Normal DPD activity and "normal" risk for fluoropyrimidine toxicity	Based on genotype, there is no indication to change dose or therapy. Use label-recommended dosage and administration.
capecitabine	The guideline does not provide a description of the impact of a DPYD activity score of 2 on capecitabine.	The guideline does not provide a recommendation for capecitabine in patients with a DPYD activity score of 2.
celecoxib	CYP2C9: Normal metabolism	Initiate therapy with recommended starting dose. In accordance with the prescribing information, use the lowest effective dosage for shortest duration consistent with individual patient treatment goals.
citalopram	CYP2C19: Normal metabolism	Initiate therapy with recommended starting dose
citalopram	The guideline does not provide a description of the impact of a normal metabolizer phenotype on citalopram.	The guideline does not provide a recommendation for citalopram in normal metabolizers.

clomipramine	Normal metabolism of tertiary amines,n/a	No recommendation
clomipramine	The guideline does not provide a description of the impact of a normal metabolizer phenotype on clomipramine.	The guideline does not provide a recommendation for clomipramine in normal metabolizers.
clopidogrel	CYP2C19: Normal clopidogrel active metabolite formation; normal on-treatment platelet reactivity	If considering clopidogrel, use at standard dose (75 mg/day) **Other Considerations** For cardiovascular indications of acute coronary syndrome (ACS) and/or percutaneous coronary intervention (PCI). ACS and/or PCI includes patients undergoing PCI for an ACS or non-ACS (elective) indication.
clopidogrel	CYP2C19: Normal clopidogrel active metabolite formation; normal on-treatment platelet reactivity	If considering clopidogrel, use at standard dose (75 mg/day) **Other Considerations** For non-acute coronary syndrome (non-ACS) and non-percutaneous coronary intervention (non-PCI) cardiovascular indications. Non-ACS, non-PCI cardiovascular indications include peripheral arterial disease and stable coronary artery disease following a recent myocardial infarction outside the setting of PCI.
clopidogrel	CYP2C19: Normal clopidogrel active metabolite formation; normal on-treatment platelet reactivity	If considering clopidogrel, use at standard dose (75 mg/day) **Other Considerations** For neurovascular indications. Neurovascular disease includes acute ischemic stroke or transient ischemic attack, secondary prevention of stroke, or prevention of thromboembolic events following neurointerventional procedures such as carotid artery stenting and stent-assisted coiling of intracranial aneurysms.
clopidogrel	The guideline does not provide a description of the impact of a normal metabolizer phenotype on clopidogrel.	The guideline does not provide a recommendation for clopidogrel in normal metabolizers.
codeine	CYP2D6: n/a	No recommendation
dapsone	G6PD: Low risk of acute hemolytic anemia	No reason to avoid based on G6PD status
desipramine	CYP2D6: n/a	No recommendation
doxepin	Normal metabolism of tertiary amines,n/a	No recommendation

efavirenz	CYP2B6: Normal efavirenz metabolism	Initiate efavirenz with standard dosing (600 mg/day) **Other Considerations** The ENCORE study showed that in treatment-naïve patients randomized to initiate efavirenz-based regimens (combined with tenofovir and emtricitabine), 400 mg/day was non-inferior to 600 mg/day regardless of CYP2B6 genotype (PMID 24522178).
efavirenz	The guideline does not provide a description of the impact of a normal metabolizer phenotype on efavirenz.	The guideline does not provide a recommendation for efavirenz in normal metabolizers.
escitalopram	CYP2C19: Normal metabolism	Initiate therapy with recommended starting dose
escitalopram	The guideline does not provide a description of the impact of a normal metabolizer phenotype on escitalopram.	The guideline does not provide a recommendation for escitalopram in normal metabolizers.
flucytosine	The guideline does not provide a description of the impact of a DPYD activity score of 2 on flucytosine.	The guideline does not provide a recommendation for flucytosine in patients with a DPYD activity score of 2.
fluorouracil	DPYD: Normal DPD activity and "normal" risk for fluoropyrimidine toxicity	Based on genotype, there is no indication to change dose or therapy. Use label-recommended dosage and administration.
fluorouracil	The guideline does not provide a description of the impact of a DPYD activity score of 2 on fluorouracil.	The guideline does not provide a recommendation for fluorouracil in patients with a DPYD activity score of 2.
flurbiprofen	CYP2C9: Normal metabolism	Initiate therapy with recommended starting dose. In accordance with the prescribing information, use the lowest effective dosage for shortest duration consistent with individual patient treatment goals.
fluvastatin	Normal exposure.,Typical myopathy risk and statin exposure.	Prescribe desired starting dose and adjust doses of fluvastatin based on disease-specific guidelines. **Other Considerations** The potential for drug-drug interactions and dose limits based on renal and hepatic function and ancestry should be evaluated prior to initiating a statin.
fluvoxamine	CYP2D6: n/a	No recommendation
fosphenytoin	Normal phenytoin metabolism,n/a	No adjustments needed from typical dosing strategies. Subsequent doses should be adjusted according to therapeutic drug monitoring, response, and side effects. An HLA-B*15:02 negative test does not eliminate the risk of phenytoin-induced SJS/TEN and patients should be carefully monitored according to a usual standard.

fosphenytoin	Normal phenytoin metabolism,n/a	No adjustments needed from typical dosing strategies. Subsequent doses should be adjusted according to therapeutic drug monitoring, response, and side effects. An HLA-B*15:02 negative test does not eliminate the risk of phenytoin-induced SJS/TEN and patients should be carefully monitored according to a usual standard.
hormonal contraceptives for systemic use	The guideline does not provide a description of the impact of the absence of Factor V Leiden on estrogen-containing contraceptives.	The guideline does not provide a recommendation for estrogen-containing contraceptives in patients who do not have the Factor V Leiden variant.
hydrocodone	CYP2D6: n/a	No recommendation
ibuprofen	CYP2C9: Normal metabolism	Initiate therapy with recommended starting dose. In accordance with the prescribing information, use the lowest effective dosage for shortest duration consistent with individual patient treatment goals.
imipramine	Normal metabolism of tertiary amines,n/a	No recommendation
imipramine	The guideline does not provide a description of the impact of a normal metabolizer phenotype on imipramine.	The guideline does not provide a recommendation for imipramine in normal metabolizers.
irinotecan	This genetic variation (IM) is more common in Western populations than the wild-type (*1/*1). This means that treatment is largely geared to patients with this genetic variation. Adjustment of the treatment is therefore not useful.	NO action is needed for this gene-drug interaction.
lornoxicam	CYP2C9: Normal metabolism	Initiate therapy with recommended starting dose. In accordance with the prescribing information, use the lowest effective dosage for shortest duration consistent with individual patient treatment goals.
lovastatin	SLCO1B1: Typical myopathy risk and statin exposure	Prescribe desired starting dose and adjust doses based on disease-specific guidelines. **Other Considerations** The potential for drug-drug interactions and dose limits based on renal and hepatic function and ancestry should be evaluated prior to initiating a statin.
meloxicam	CYP2C9: Normal metabolism	Initiate therapy with recommended starting dose. In accordance with the prescribing information, use the lowest effective dosage for shortest duration consistent with individual patient treatment goals.
methylene blue	G6PD: Low risk of acute hemolytic anemia	No reason to avoid based on G6PD status
nitrofurantoin	G6PD: Low risk of acute hemolytic anemia	No reason to avoid based on G6PD status

nortriptyline	CYP2D6: n/a	No recommendation **Other Considerations** Because CYP2D6 phenotype could not be assigned based on genotyping performed, therapuetic monitoring should be considered. If therapuetic monitoring cannot be performed, monitor closely for toxicity and/or efficacy.
ondansetron	CYP2D6: n/a	No recommendation
paroxetine	CYP2D6: n/a	No recommendation
pegloticase	G6PD: Low risk of acute hemolytic anemia	No reason to avoid based on G6PD status
phenytoin	Normal phenytoin metabolism,n/a	No adjustments needed from typical dosing strategies. Subsequent doses should be adjusted according to therapeutic drug monitoring, response, and side effects. An HLA-B*15:02 negative test does not eliminate the risk of phenytoin-induced SJS/TEN and patients should be carefully monitored according to a usual standard.
phenytoin	Normal phenytoin metabolism,n/a	No adjustments needed from typical dosing strategies. Subsequent doses should be adjusted according to therapeutic drug monitoring, response, and side effects. An HLA-B*15:02 negative test does not eliminate the risk of phenytoin-induced SJS/TEN and patients should be carefully monitored according to a usual standard.
phenytoin	The guideline does not provide a description of the impact of a normal metabolizer phenotype on phenytoin.	The guideline does not provide a recommendation for phenytoin in normal metabolizers.
piroxicam	CYP2C9: Normal metabolism	Initiate therapy with recommended starting dose. In accordance with the prescribing information, use the lowest effective dosage for shortest duration consistent with individual patient treatment goals.
pitavastatin	SLCO1B1: Typical myopathy risk and statin exposure	Prescribe desired starting dose and adjust doses based on disease-specific guidelines. **Other Considerations** The potential for drug-drug interactions and dose limits based on renal and hepatic function and ancestry should be evaluated prior to initiating a statin.
pravastatin	SLCO1B1: Typical myopathy risk and statin exposure	Prescribe desired starting dose and adjust doses based on disease-specific guidelines. **Other Considerations** The potential for drug-drug interactions and dose limits based on renal and hepatic function and ancestry should be evaluated prior to initiating a statin.

primaquine	G6PD: Low risk of acute hemolytic anemia	No reason to avoid based on G6PD status
quetiapine	The guideline does not provide a description of the impact of a normal metabolizer phenotype on quetiapine.	The guideline does not provide a recommendation for quetiapine in normal metabolizers.
rasburicase	G6PD: Low risk of acute hemolytic anemia	No reason to avoid based on G6PD status
rosuvastatin	Typical myopathy risk and rosuvastatin exposure,Typical myopathy risk and statin exposure	Prescribe desired starting dose and adjust doses of rosuvastatin based on disease-specific and specific population guidelines. **Other Considerations** The potential for drug-drug interactions and dose limits based on renal and hepatic function and Asian ancestry should be evaluated prior to initiating rosuvastatin.
sertraline	Normal metabolism of sertraline to less active compounds.,Normal metabolism	Initiate therapy with recommended starting dose.
sertraline	The guideline does not provide a description of the impact of a normal metabolizer phenotype on sertraline.	The guideline does not provide a recommendation for sertraline in normal metabolizers.
simvastatin	SLCO1B1: Typical myopathy risk and statin exposure	Prescribe desired starting dose and adjust doses based on disease-specific guidelines. **Other Considerations** The potential for drug-drug interactions and dose limits based on renal and hepatic function and ancestry should be evaluated prior to initiating a statin.
siponimod	The guideline does not provide a description of the impact of a normal metabolizer phenotype on siponimod.	The guideline does not provide a recommendation for siponimod in normal metabolizers.
tacrolimus	CYP3A5: Higher ("normal") dose-adjusted trough concentrations of tacrolimus and increased chance of achieving target tacrolimus concentrations.	Initiate therapy with standard recommended dose. Use therapeutic drug monitoring to guide dose adjustments. **Other Considerations** This recommendation includes the use of tacrolimus in kidney, heart, lung and hematopoietic stem cell transplant patients, and liver transplant patients where the donor and recipient genotypes are identical. Typically with other CYP enzymes, a normal metabolizer would be classified as having normal metabolism, and therefore, the drug dose would not change based on the patient's genotype. However, in the case of CYP3A5 and tacrolimus, a CYP3A5 expresser (i.e., CYP3A5 normal metabolizer or intermediate metabolizer) would require a higher recommended starting dose and the CYP3A5 non-expresser (i.e., poor metabolizer) would require the standard recommended starting dose.

tacrolimus	The guideline does not provide a description of the impact of a non-expressor phenotype on tacrolimus.	The guideline does not provide a recommendation for tacrolimus in CYP3A5 non-expressers.
tafenoquine	G6PD: Low risk of acute hemolytic anemia	No reason to avoid based on G6PD status **Other Considerations** Tafenoquine's safety has been established for a G6PD enzyme activity ≥70% of normal. (Inclusion criteria for clinical trials involving tafenoquine included G6PD activity ≥70%.)
tamoxifen	CYP2D6: n/a	No recommendation
tegafur	The guideline does not provide a description of the impact of a DPYD activity score of 2 on tegafur.	The guideline does not provide a recommendation for tegafur in patients with a DPYD activity score of 2.
tenoxicam	CYP2C9: Normal metabolism	Initiate therapy with recommended starting dose. In accordance with the prescribing information, use the lowest effective dosage for shortest duration consistent with individual patient treatment goals.
toluidine blue	G6PD: Low risk of acute hemolytic anemia	No reason to avoid based on G6PD status **Other Considerations** Toluidine blue classification strength is based on extrapolation from methylene blue data
tramadol	CYP2D6: n/a	No recommendation
trimipramine	Normal metabolism of tertiary amines,n/a	No recommendation
tropisetron	CYP2D6: n/a	No recommendation
venlafaxine	CYP2D6: n/a	No recommendation
voriconazole	CYP2C19: Normal voriconazole metabolism	Initiate therapy with recommended standard of care dosing **Other Considerations** Further dose adjustments or selection of alternative therapy may be necessary due to other clinical factors, such as drug interactions, hepatic function, renal function, species, site of infection, therapeutic drug monitoring, and comorbidities.
voriconazole	CYP2C19: Normal voriconazole metabolism	Initiate therapy with recommended standard of care dosing **Other Considerations** Further dose adjustments or selection of alternative therapy may be necessary due to other clinical factors, such as drug interactions, hepatic function, renal function, species, site of infection, therapeutic drug monitoring, and comorbidities.
voriconazole	The guideline does not provide a description of the impact of a normal metabolizer phenotype on voriconazole.	The guideline does not provide a recommendation for voriconazole in normal metabolizers.

vortioxetine	CYP2D6: n/a	No recommendation
azathioprine	Grade >=2 leukopaenia occurs in 96% of these patients with standard therapy. The genetic variation increases the concentration of the fully activated metabolite of azathioprine and mercaptopurine.	Avoid azathioprine and mercaptopurine. If it is not possible to avoid azathioprine and mercaptopurine: use 10% of the standard dose and advise patients to seek medical attention when symptoms of myelosuppression (such as severe sore throat in combination with fever, regular nosebleeds and tendency to bruising) occur. Any adjustment of the initial dose should be guided by toxicity (monitoring of blood counts) and efficacy. Note: The percentage of 10% is based on the analogy with TPMT, for which the gene variants have a comparable effect on toxicity to those of NUDT15. A percentage of < 20% was calculated for NUDT15 PM, but there were insufficient data available to calculate the exact percentage. Note: Dose adjustment based on the total of 6-TGN metabolites is not possible for these patients, as they develop toxicity within the therapeutic range that applies for patients without gene variants.
mercaptopurine	Grade ≥2 leukopaenia occurs in 96% of these patients with standard therapy. The genetic variation increases the concentration of the fully activated metabolite of azathioprine and mercaptopurine.	Avoid azathioprine and mercaptopurine. If it is not possible to avoid azathioprine and mercaptopurine: use 10% of the standard dose and advise patients to seek medical attention when symptoms of myelosuppression (such as severe sore throat in combination with fever, regular nosebleeds and tendency to bruising) occur. Any adjustment of the initial dose should be guided by toxicity (monitoring of blood counts) and efficacy. Note: The percentage of 10% is based on the analogy with TPMT, for which the gene variants have a comparable effect on toxicity to those of NUDT15. A percentage of < 20% was calculated for NUDT15 PM, but there were insufficient data available to calculate the exact percentage. Note: Dose adjustment based on the total of 6-TGN metabolites is not possible for these patients, as they develop toxicity within the therapeutic range that applies for patients without gene variants.

● **thioguanine**	Grade 2 leukopaenia occurs in an estimated 95% of these patients with standard therapy. The genetic variation increases the concentration of the fully activated metabolite of thioguanine.	Avoid thioguanine. If it is not possible to avoid tioguanine: use 10% of the standard dose and advise patients to seek medical attention when symptoms of myelosuppression (such as severe sore throat in combination with fever, regular nosebleeds and tendency to bruising) occur. Any adjustment of the initial dose should be guided by toxicity (monitoring of blood counts) and efficacy. Monitoring should be performed at an increased frequency. NOTE: The percentage of 10% is based on the analogy with azathioprine and mercaptopurine and the analogy with TPMT, for which the gene variants have a comparable effect on toxicity to those of NUDT15. For NUDT15 PM, a percentage of < 20% was calculated for azathioprine and mercaptopurine, but there were insufficient data available to calculate the exact percentage. NOTE: Dose adjustment based on the total of 6-TGN metabolites is not possible for these patients, as they develop toxicity within the therapeutic range that applies for patients without gene variants.

Supplementary Details

1. Pharmacogenomic Guidance Overview

	Standard Precautions	Use with Cautions	Consider Alternatives
Pulmonary			ivacaftor
Cardiology	atorvastatin clopidogrel fluvastatin lovastatin pitavastatin pravastatin rosuvastatin simvastatin	acenocoumarol phenprocoumon	
Rheumatology	celecoxib flurbiprofen ibuprofen lornoxicam pegloticase piroxicam tenoxicam	azathioprine	
Gastroenterology	ondansetron	dexlansoprazole lansoprazole omeprazole pantoprazole	
Oncology	allopurinol capecitabine fluorouracil irinotecan rasburicase tamoxifen tegafur tropisetron	mercaptopurine thioguanine	
Hematology	warfarin methylene blue		
Infectious Disease	atazanavir efavirenz nitrofurantoin primaquine tafenoquine voriconazole		

	Standard Precautions	Use with Cautions	Consider Alternatives
Anesthesiology	desflurane enflurane halothane isoflurane methoxyflurane sevoflurane succinylcholine codeine meloxicam tramadol		
Psychiatry	amitriptyline atomoxetine citalopram clomipramine desipramine doxepin escitalopram fluvoxamine imipramine nortriptyline paroxetine phenytoin quetiapine sertraline trimipramine venlafaxine vortioxetine		
Dermatology	dapsone		
Infection	flucytosine		
Neurology	fosphenytoin siponimod		
Gynecology	hormonal contraceptives for systemic use		
Pain	hydrocodone		
Transplantation	tacrolimus		
Others	toluidine blue		

2. Detailed Prescribing Guidance

The prescribing recommendations are based on the information provided to the software. For a detailed disclaimer see Disclaimer & Information.

NOVOCLINIC

Pharmacogenomics Report

John Doe (4376)

ivacaftor

CPIC

Population: General **Classification:** Moderate

Implications:

CFTR:Reference/Reference
CFTR: An individual diagnosed with cystic fibrosis (CF) and negative for a CFTR variant listed in the FDA-approved drug label as being responsive to ivacaftor.

Recommendation: Ivacaftor is not recommended

Citation:

- Clinical Pharmacogenetics Implementation Consortium (CPIC) guidelines for ivacaftor therapy in the context of CFTR genotype. Clinical pharmacology and therapeutics. 2014. PMID:24598717

acenocoumarol

DPWG

Population: Unspecified **Classification:** N/A

Implications:

VKORC1:rs9923231 variant (T)/rs9923231 variant (T)
An INR ≥ 6, resulting in an increased risk of bleeding, occurs in 8-12% of these patients during the first weeks of treatment with standard regulation by the Anticoagulation Clinic. The genetic variation increases the sensitivity to acenocoumarol.

Recommendation: Monitoring by the ANTICOAGULATION CLINIC (National INR Monitoring Service):

- recommend to use 50% of the standard initial dose

OTHERWISE:

- recommend to use 50% of the standard initial dose
- recommend more frequent monitoring of the INR

Citation:

- Pharmacogenetics: from bench to byte--an update of guidelines. Clinical pharmacology and therapeutics. 2011. PMID:21412232

Pharmacogenomics Report

John Doe (4376)

azathioprine

CPIC

Population: General **Classification:** Strong

Implications:

NUDT15:*2/*6,TPMT:*1/*1

NUDT15: Increased risk of thiopurine-related leukopenia, neutropenia, myelosuppression,TPMT: Lower concentrations of TGN metabolites, higher MeTIMP, this is the 'normal' pattern. Normal risk of thiopurine-related leukopenia, neutropenia, myelosuppression.

Recommendation:

Start with reduced starting doses (30%-80% of normal dose) if normal starting dose is 2-3 mg/kg/day (e.g., 0.6 – 2.4 mg/kg/day), and adjust doses of azathioprine based on degree of myelosuppression and disease-specific guidelines. Allow 2-4 weeks to reach steady-state after each dose adjustment (PMID 20354201, 11302950, 15606506, 16530532).

Other Considerations

Normal starting doses vary by race/ethnicity and treatment regimens. If standard dose is below normal recommended dose, dose reduction might not be recommended for intermediate metabolizers.

DPWG

Population: Unspecified **Classification:** N/A

Implications:

NUDT15:*2/*6

Grade >=2 leukopaenia occurs in 96% of these patients with standard therapy. The genetic variation increases the concentration of the fully activated metabolite of azathioprine and mercaptopurine.

Recommendation:

Avoid azathioprine and mercaptopurine. If it is not possible to avoid azathioprine and mercaptopurine: use 10% of the standard dose and advise patients to seek medical attention when symptoms of myelosuppression (such as severe sore throat in combination with fever, regular nosebleeds and tendency to bruising) occur. Any adjustment of the initial dose should be guided by toxicity (monitoring of blood counts) and efficacy. Note: The percentage of 10% is based on the analogy with TPMT, for which the gene variants have a comparable effect on toxicity to those of NUDT15. A percentage of < 20% was calculated for NUDT15 PM, but there were insufficient data available to calculate the exact percentage. Note: Dose adjustment based on the total of 6-TGN metabolites is not possible for these patients, as they develop toxicity within the therapeutic range that applies for patients without gene variants.

DPWG

Population: Unspecified **Classification:** No recommendation

Implications:

TPMT:*1/*1

The guideline does not provide a description of the impact of a normal metabolizer phenotype on azathioprine.

Recommendation:

The guideline does not provide a recommendation for azathioprine in normal metabolizers.

Citation:

- Clinical Pharmacogenetics Implementation Consortium guidelines for thiopurine methyltransferase genotype and thiopurine dosing. Clinical pharmacology and therapeutics. 2011. PMID:21270794
- Clinical pharmacogenetics implementation consortium guidelines for thiopurine methyltransferase genotype and thiopurine dosing: 2013 update. Clinical pharmacology and therapeutics. 2013. PMID:23422873
- Clinical Pharmacogenetics Implementation Consortium Guideline for Thiopurine Dosing Based on TPMT and NUDT15 Genotypes: 2018 Update. Clinical pharmacology and therapeutics. 2019. PMID:30447069
- Pharmacogenetics: from bench to byte--an update of guidelines. Clinical pharmacology and therapeutics. 2011. PMID:21412232

NOVOCLINIC

Pharmacogenomics Report

John Doe (4376)

dexlansoprazole

CPIC

Population: General **Classification:** Optional

Implications: **CYP2C19:*1/*1**
CYP2C19: Normal PPI metabolism; may be at increased risk of therapeutic failure compared to CYP2C19 IMs and PMs

Recommendation: Initiate standard starting daily dose. Consider increasing dose by 50-100% for the treatment of H. pylori infection and erosive esophagitis. Daily dose may be given in divided doses. Monitor for efficacy.

Citation:

- Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for CYP2C19 and Proton Pump Inhibitor Dosing. Clinical pharmacology and therapeutics. 2021. PMID:32770672

lansoprazole

CPIC

Population: General **Classification:** Moderate

Implications: **CYP2C19:*1/*1**
CYP2C19: Normal PPI metabolism; may be at increased risk of therapeutic failure compared to CYP2C19 IMs and PMs

Recommendation: Initiate standard starting daily dose. Consider increasing dose by 50-100% for the treatment of H. pylori infection and erosive esophagitis. Daily dose may be given in divided doses. Monitor for efficacy.

DPWG

Population: Unspecified **Classification:** No recommendation

Implications: **CYP2C19:*1/*1**
The guideline does not provide a description of the impact of a normal metabolizer phenotype on lansoprazole.

Recommendation: The guideline does not provide a recommendation for lansoprazole in normal metabolizers.

Citation:

- Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for CYP2C19 and Proton Pump Inhibitor Dosing. Clinical pharmacology and therapeutics. 2021. PMID:32770672
- Pharmacogenetics: from bench to byte--an update of guidelines. Clinical pharmacology and therapeutics. 2011. PMID:21412232

Pharmacogenomics Report

John Doe (4376)

mercaptopurine

CPIC

Population: General **Classification:** Strong

Implications:

NUDT15:*2/*6,TPMT:*1/*1

NUDT15: Increased risk of thiopurine-related leukopenia, neutropenia, myelosuppression,TPMT: Lower concentrations of TGN metabolites, higher MeTIMP, this is the 'normal' pattern. Normal risk of thiopurine-related leukopenia, neutropenia, myelosuppression.

Recommendation:

Start with reduced starting doses (30%-80% of normal dose) if normal starting dose is ≥75 mg/m2/day or ≥ 1.5 mg/kg/day (e.g., start at 22.5-60 mg/m2/day or 0.45-1.2 mg/kg/day) and adjust doses of mercaptopurine based on degree of myelosuppression and disease-specific guidelines. Allow 2-4 weeks to reach steady-state after each dose adjustment. If myelosuppression occurs, and depending on other therapy, emphasis should be on reducing mercaptopurine over other agents (PMID 20354201, 18685564, 8857546, 18987654, 20010622, 16401827, 11302950, 16530532, 9634537). If normal starting dose is already < 75 mg/m2/day or < 1.5 mg/kg/day, dose reduction may not be recommended.

Other Considerations

Normal starting doses vary by race/ethnicity and treatment regimens. If standard dose is below normal recommended dose, dose reduction might not be recommended for intermediate metabolizers.

DPWG

Population: Unspecified **Classification:** N/A

Implications:

NUDT15:*2/*6

Grade ≥2 leukopaenia occurs in 96% of these patients with standard therapy. The genetic variation increases the concentration of the fully activated metabolite of azathioprine and mercaptopurine.

Recommendation:

Avoid azathioprine and mercaptopurine. If it is not possible to avoid azathioprine and mercaptopurine: use 10% of the standard dose and advise patients to seek medical attention when symptoms of myelosuppression (such as severe sore throat in combination with fever, regular nosebleeds and tendency to bruising) occur. Any adjustment of the initial dose should be guided by toxicity (monitoring of blood counts) and efficacy. Note: The percentage of 10% is based on the analogy with TPMT, for which the gene variants have a comparable effect on toxicity to those of NUDT15. A percentage of < 20% was calculated for NUDT15 PM, but there were insufficient data available to calculate the exact percentage. Note: Dose adjustment based on the total of 6-TGN metabolites is not possible for these patients, as they develop toxicity within the therapeutic range that applies for patients without gene variants.

DPWG

Population: Unspecified **Classification:** No recommendation

Implications:

TPMT:*1/*1

The guideline does not provide a description of the impact of a normal metabolizer phenotype on mercaptopurine.

Recommendation:

The guideline does not provide a recommendation for mercaptopurine in normal metabolizers.

Citation:

- Clinical Pharmacogenetics Implementation Consortium guidelines for thiopurine methyltransferase genotype and thiopurine dosing. Clinical pharmacology and therapeutics. 2011. PMID:21270794
- Clinical pharmacogenetics implementation consortium guidelines for thiopurine methyltransferase genotype and thiopurine dosing: 2013 update. Clinical pharmacology and therapeutics. 2013. PMID:23422873
- Clinical Pharmacogenetics Implementation Consortium Guideline for Thiopurine Dosing Based on TPMT and NUDT15 Genotypes: 2018 Update. Clinical pharmacology and therapeutics. 2019. PMID:30447069
- Pharmacogenetics: from bench to byte--an update of guidelines. Clinical pharmacology and therapeutics. 2011. PMID:21412232

Pharmacogenomics Report

John Doe (4376)

omeprazole

CPIC

Population: General **Classification:** Moderate

Implications:

CYP2C19:*1/*1

CYP2C19: Normal PPI metabolism; may be at increased risk of therapeutic failure compared to CYP2C19 IMs and PMs

Recommendation: Initiate standard starting daily dose. Consider increasing dose by 50-100% for the treatment of H. pylori infection and erosive esophagitis. Daily dose may be given in divided doses. Monitor for efficacy.

DPWG

Population: Unspecified **Classification:** No recommendation

Implications:

CYP2C19:*1/*1

The guideline does not provide a description of the impact of a normal metabolizer phenotype on omeprazole.

Recommendation: The guideline does not provide a recommendation for omeprazole in normal metabolizers.

Citation:

- Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for CYP2C19 and Proton Pump Inhibitor Dosing. Clinical pharmacology and therapeutics. 2021. PMID:32770672
- Pharmacogenetics: from bench to byte--an update of guidelines. Clinical pharmacology and therapeutics. 2011. PMID:21412232

pantoprazole

CPIC

Population: General **Classification:** Moderate

Implications:

CYP2C19:*1/*1

CYP2C19: Normal PPI metabolism; may be at increased risk of therapeutic failure compared to CYP2C19 IMs and PMs

Recommendation: Initiate standard starting daily dose. Consider increasing dose by 50-100% for the treatment of H. pylori infection and erosive esophagitis. Daily dose may be given in divided doses. Monitor for efficacy.

DPWG

Population: Unspecified **Classification:** No recommendation

Implications:

CYP2C19:*1/*1

The guideline does not provide a description of the impact of a normal metabolizer phenotype on pantoprazole.

Recommendation: The guideline does not provide a recommendation for pantoprazole in normal metabolizers.

Citation:

- Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for CYP2C19 and Proton Pump Inhibitor Dosing. Clinical pharmacology and therapeutics. 2021. PMID:32770672
- Pharmacogenetics: from bench to byte--an update of guidelines. Clinical pharmacology and therapeutics. 2011. PMID:21412232

Pharmacogenomics Report

John Doe (4376)

phenprocoumon

DPWG

Population: Unspecified **Classification:** N/A

Implications:

VKORC1:rs9923231 variant (T)/rs9923231 variant (T)

An INR ≥ 6, resulting in an increased risk of bleeding, occurs in 17% of these patients with standard regulation by the Anticoagulation Clinic. The genetic variation increases the sensitivity to phenprocoumon.

Recommendation:

Monitoring by a ANTICOAGULATION CLINIC:

- recommend to use 50% of the standard initial dose

NO monitoring by a anticoagulation clinic:

- recommend to use 50% of the standard initial dose
- recommend more frequent monitoring of the INR

Citation:

- Pharmacogenetics: from bench to byte--an update of guidelines. Clinical pharmacology and therapeutics. 2011. PMID:21412232

thioguanine

CPIC

Population: General **Classification:** Moderate

Implications:

NUDT15:*2/*6,TPMT:*1/*1

NUDT15: Increased risk of thiopurine-related leukopenia, neutropenia, myelosuppression,TPMT: Lower concentrations of TGN metabolites, but note that TGN after thioguanine are 5-10X higher than TGN after mercaptopurine or azathioprine. Normal risk of thiopurine-related leukopenia, neutropenia, myelosuppression.

Recommendation:

Start with reduced doses (50% to 80% of normal dose) if normal starting dose is ≥ 40-60 mg/m2/day (e.g., 20-48 mg/m2/day) and adjust doses of thioguanine based on degree of myelosuppression and disease-specific guidelines. Allow 2-4 weeks to reach steady-state after each dose adjustment. If myelosuppression occurs, and depending on other therapy, emphasis should be on reducing thioguanine over other agents (PMID 20354201, 11037857).

Other Considerations

Normal starting doses vary by race/ethnicity and treatment regimens. If standard dose is below normal recommended dose, dose reduction might not be recommended for intermediate metabolizers.

DPWG

Population: Unspecified **Classification:** N/A

Implications:

NUDT15:*2/*6

Grade 2 leukopaenia occurs in an estimated 95% of these patients with standard therapy. The genetic variation increases the concentration of the fully activated metabolite of thioguanine.

Recommendation:

Avoid thioguanine. If it is not possible to avoid tioguanine: use 10% of the standard dose and advise patients to seek medical attention when symptoms of myelosuppression (such as severe sore throat in combination with fever, regular nosebleeds and tendency to bruising) occur. Any adjustment of the initial dose should be guided by toxicity (monitoring of blood counts) and efficacy. Monitoring should be performed at an increased frequency. NOTE: The percentage of 10% is based on the analogy with azathioprine and mercaptopurine and the analogy with TPMT, for which the gene variants have a comparable effect on toxicity to those of NUDT15. For NUDT15 PM, a percentage of < 20% was calculated for azathioprine and mercaptopurine, but there were insufficient data available to calculate the exact percentage. NOTE: Dose adjustment based on the total of 6-TGN metabolites is not possible for these patients, as they develop toxicity within the therapeutic range that applies for patients without gene variants.

DPWG

Population: Unspecified **Classification:** No recommendation

Implications:

TPMT:*1/*1

The guideline does not provide a description of the impact of a normal metabolizer phenotype on thioguanine.

Recommendation:

The guideline does not provide a recommendation for thioguanine in normal metabolizers.

Citation:

- Clinical Pharmacogenetics Implementation Consortium guidelines for thiopurine methyltransferase genotype and thiopurine dosing. Clinical pharmacology and therapeutics. 2011. PMID:21270794
- Clinical pharmacogenetics implementation consortium guidelines for thiopurine methyltransferase genotype and thiopurine dosing: 2013 update. Clinical pharmacology and therapeutics. 2013. PMID:23422873
- Clinical Pharmacogenetics Implementation Consortium Guideline for Thiopurine Dosing Based on TPMT and NUDT15 Genotypes: 2018 Update. Clinical pharmacology and therapeutics. 2019. PMID:30447069
- Pharmacogenetics: from bench to byte--an update of guidelines. Clinical pharmacology and therapeutics. 2011. PMID:21412232

Pharmacogenomics Report

John Doe (4376)

warfarin

CPIC

Population: N/A **Classification:** N/A

Implications: **CYP2C9:*1/*1,CYP4F2:*1/*1,VKORC1:rs9923231 variant (T)/rs9923231 variant (T)**
N/A

Recommendation: N/A

DPWG

Population: Unspecified **Classification:** No recommendation

Implications: **CYP2C9:*1/*1**
The guideline does not provide a description of the impact of a normal metabolizer phenotype on warfarin.

Recommendation: The guideline does not provide a recommendation for warfarin in normal metabolizers.

DPWG

Population: Unspecified **Classification:** N/A

Implications: **VKORC1:rs9923231 variant (T)/rs9923231 variant (T)**
The genetic variation results in increased sensitivity to warfarin. This results in an increase in the risk of excessively severe inhibition of blood clotting (INR > 4) during the first month of the treatment.

Recommendation: Use 60% of the standard initial dose. The genotype-specific initial dose and maintenance dose can be calculated using an algorithm, as used in EU-PACT: see https://www.knmp.nl/patientenzorg/medicatiebewaking/farmacogenetica. From day 6 on the standard algorithm without genotype information can be used to calculate the dose.

Citation:

- Clinical Pharmacogenetics Implementation Consortium Guidelines for CYP2C9 and VKORC1 genotypes and warfarin dosing. Clinical pharmacology and therapeutics. 2011. PMID:21900891
- Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for Pharmacogenetics-Guided Warfarin Dosing: 2017 Update. Clinical pharmacology and therapeutics. 2017. PMID:28198005

Pharmacogenomics Report

John Doe (4376)

atazanavir

CPIC

Population: General **Classification:** Strong

Implications:

UGT1A1:*1/*6

UGT1A1: Somewhat decreased UGT1A1 activity; low likelihood of bilirubin-related discontinuation of atazanavir.

Recommendation: There is no need to avoid prescribing of atazanavir based on UGT1A1 genetic test result. Inform the patient that some patients stop atazanavir because of jaundice (yellow eyes and skin), but that this patient's genotype makes this unlikely (less than about a 1 in 20 chance of stopping atazanavir because of jaundice).

Other Considerations

All studies correlating UGT1A1 genotypes with atazanavir adverse events have involved ritonavir boosting. However, concentration-time profiles are equivalent when boosted with either cobicistat or ritonavir (PMID 23532097), and bilirubin-related adverse events including discontinuation of atazanavir occur in a similar percentage of patients prescribed atazanavir with cobicistat or ritonavir (PMID 23532097). Associations between UGT1A1 genotype, bilirubin elevations, and atazanavir/r discontinuation therefore almost certainly translate to atazanavir/cobicistat.

Citation:

- Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for UGT1A1 and Atazanavir Prescribing. Clinical pharmacology and therapeutics. 2016. PMID:26417955

desflurane

CPIC

Population: General **Classification:** Strong

Implications:

CACNA1S:Reference/Reference

These results do not eliminate the chance that this patient is susceptible to Malignant Hyperthermia. The genetic cause of about half of all MH survivors, with MH susceptibility confirmed by contracture test, remains unknown (PMID: 28902675).

Recommendation: Clinical findings, family history, further genetic testing and other laboratory data should guide use of halogenated volatile anesthetics or depolarizing muscle relaxants.

CPIC

Population: General **Classification:** Strong

Implications:

RYR1:Reference/Reference

These results do not eliminate the chance that this patient is susceptible to Malignant Hyperthermia. The genetic cause of about half of all MH survivors, with MH susceptibility confirmed by contracture test, remains unknown (PMID: 28902675). A negative or inconclusive genetic test cannot be assumed to indicate normal RYR1-related phenotype and should be interpreted in context of clinical findings, family history and other laboratory data.

Recommendation: Clinical findings, family history, further genetic testing and other laboratory data should guide use of halogenated volatile anesthetics or depolarizing muscle relaxants.

Citation:

- Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for the Use of Potent Volatile Anesthetic Agents and Succinylcholine in the Context of RYR1 or CACNA1S Genotypes. Clinical pharmacology and therapeutics. 2019. PMID:30499100

Pharmacogenomics Report

John Doe (4376)

enflurane

CPIC

Population: General **Classification:** Strong

Implications: **CACNA1S:Reference/Reference**
These results do not eliminate the chance that this patient is susceptible to Malignant Hyperthermia. The genetic cause of about half of all MH survivors, with MH susceptibility confirmed by contracture test, remains unknown (PMID: 28902675).

Recommendation: Clinical findings, family history, further genetic testing and other laboratory data should guide use of halogenated volatile anesthetics or depolarizing muscle relaxants.

CPIC

Population: General **Classification:** Strong

Implications: **RYR1:Reference/Reference**
These results do not eliminate the chance that this patient is susceptible to Malignant Hyperthermia. The genetic cause of about half of all MH survivors, with MH susceptibility confirmed by contracture test, remains unknown (PMID: 28902675). A negative or inconclusive genetic test cannot be assumed to indicate normal RYR1-related phenotype and should be interpreted in context of clinical findings, family history and other laboratory data.

Recommendation: Clinical findings, family history, further genetic testing and other laboratory data should guide use of halogenated volatile anesthetics or depolarizing muscle relaxants.

Citation:

- Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for the Use of Potent Volatile Anesthetic Agents and Succinylcholine in the Context of RYR1 or CACNA1S Genotypes. Clinical pharmacology and therapeutics. 2019. PMID:30499100

Pharmacogenomics Report

John Doe (4376)

halothane

CPIC

Population: General **Classification:** Strong

Implications: **CACNA1S:Reference/Reference**
These results do not eliminate the chance that this patient is susceptible to Malignant Hyperthermia. The genetic cause of about half of all MH survivors, with MH susceptibility confirmed by contracture test, remains unknown (PMID: 28902675).

Recommendation: Clinical findings, family history, further genetic testing and other laboratory data should guide use of halogenated volatile anesthetics or depolarizing muscle relaxants.

CPIC

Population: General **Classification:** Strong

Implications: **RYR1:Reference/Reference**
These results do not eliminate the chance that this patient is susceptible to Malignant Hyperthermia. The genetic cause of about half of all MH survivors, with MH susceptibility confirmed by contracture test, remains unknown (PMID: 28902675). A negative or inconclusive genetic test cannot be assumed to indicate normal RYR1-related phenotype and should be interpreted in context of clinical findings, family history and other laboratory data.

Recommendation: Clinical findings, family history, further genetic testing and other laboratory data should guide use of halogenated volatile anesthetics or depolarizing muscle relaxants.

Citation:

- Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for the Use of Potent Volatile Anesthetic Agents and Succinylcholine in the Context of RYR1 or CACNA1S Genotypes. Clinical pharmacology and therapeutics. 2019. PMID:30499100

Pharmacogenomics Report

John Doe (4376)

isoflurane

CPIC

Population: General **Classification:** Strong

Implications: **CACNA1S:Reference/Reference**
These results do not eliminate the chance that this patient is susceptible to Malignant Hyperthermia. The genetic cause of about half of all MH survivors, with MH susceptibility confirmed by contracture test, remains unknown (PMID: 28902675).

Recommendation: Clinical findings, family history, further genetic testing and other laboratory data should guide use of halogenated volatile anesthetics or depolarizing muscle relaxants.

CPIC

Population: General **Classification:** Strong

Implications: **RYR1:Reference/Reference**
These results do not eliminate the chance that this patient is susceptible to Malignant Hyperthermia. The genetic cause of about half of all MH survivors, with MH susceptibility confirmed by contracture test, remains unknown (PMID: 28902675). A negative or inconclusive genetic test cannot be assumed to indicate normal RYR1-related phenotype and should be interpreted in context of clinical findings, family history and other laboratory data.

Recommendation: Clinical findings, family history, further genetic testing and other laboratory data should guide use of halogenated volatile anesthetics or depolarizing muscle relaxants.

Citation:

- Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for the Use of Potent Volatile Anesthetic Agents and Succinylcholine in the Context of RYR1 or CACNA1S Genotypes. Clinical pharmacology and therapeutics. 2019. PMID:30499100

methoxyflurane

CPIC

Population: General **Classification:** Strong

Implications: **CACNA1S:Reference/Reference**
These results do not eliminate the chance that this patient is susceptible to Malignant Hyperthermia. The genetic cause of about half of all MH survivors, with MH susceptibility confirmed by contracture test, remains unknown (PMID: 28902675).

Recommendation: Clinical findings, family history, further genetic testing and other laboratory data should guide use of halogenated volatile anesthetics or depolarizing muscle relaxants.

CPIC

Population: General **Classification:** Strong

Implications: **RYR1:Reference/Reference**
These results do not eliminate the chance that this patient is susceptible to Malignant Hyperthermia. The genetic cause of about half of all MH survivors, with MH susceptibility confirmed by contracture test, remains unknown (PMID: 28902675). A negative or inconclusive genetic test cannot be assumed to indicate normal RYR1-related phenotype and should be interpreted in context of clinical findings, family history and other laboratory data.

Recommendation: Clinical findings, family history, further genetic testing and other laboratory data should guide use of halogenated volatile anesthetics or depolarizing muscle relaxants.

Citation:

- Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for the Use of Potent Volatile Anesthetic Agents and Succinylcholine in the Context of RYR1 or CACNA1S Genotypes. Clinical pharmacology and therapeutics. 2019. PMID:30499100

Pharmacogenomics Report

John Doe (4376)

sevoflurane

CPIC

Population: General **Classification:** Strong

Implications: **CACNA1S:Reference/Reference**
These results do not eliminate the chance that this patient is susceptible to Malignant Hyperthermia. The genetic cause of about half of all MH survivors, with MH susceptibility confirmed by contracture test, remains unknown (PMID: 28902675).

Recommendation: Clinical findings, family history, further genetic testing and other laboratory data should guide use of halogenated volatile anesthetics or depolarizing muscle relaxants.

CPIC

Population: General **Classification:** Strong

Implications: **RYR1:Reference/Reference**
These results do not eliminate the chance that this patient is susceptible to Malignant Hyperthermia. The genetic cause of about half of all MH survivors, with MH susceptibility confirmed by contracture test, remains unknown (PMID: 28902675). A negative or inconclusive genetic test cannot be assumed to indicate normal RYR1-related phenotype and should be interpreted in context of clinical findings, family history and other laboratory data.

Recommendation: Clinical findings, family history, further genetic testing and other laboratory data should guide use of halogenated volatile anesthetics or depolarizing muscle relaxants.

Citation:

- Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for the Use of Potent Volatile Anesthetic Agents and Succinylcholine in the Context of RYR1 or CACNA1S Genotypes. Clinical pharmacology and therapeutics. 2019. PMID:30499100

Pharmacogenomics Report

John Doe (4376)

succinylcholine

CPIC

Population: General **Classification:** Strong

Implications: **CACNA1S:Reference/Reference**
These results do not eliminate the chance that this patient is susceptible to Malignant Hyperthermia. The genetic cause of about half of all MH survivors, with MH susceptibility confirmed by contracture test, remains unknown (PMID: 28902675).

Recommendation: Clinical findings, family history, further genetic testing and other laboratory data should guide use of halogenated volatile anesthetics or depolarizing muscle relaxants.

CPIC

Population: General **Classification:** Strong

Implications: **RYR1:Reference/Reference**
These results do not eliminate the chance that this patient is susceptible to Malignant Hyperthermia. The genetic cause of about half of all MH survivors, with MH susceptibility confirmed by contracture test, remains unknown (PMID: 28902675). A negative or inconclusive genetic test cannot be assumed to indicate normal RYR1-related phenotype and should be interpreted in context of clinical findings, family history and other laboratory data.

Recommendation: Clinical findings, family history, further genetic testing and other laboratory data should guide use of halogenated volatile anesthetics or depolarizing muscle relaxants.

Citation:

- Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for the Use of Potent Volatile Anesthetic Agents and Succinylcholine in the Context of RYR1 or CACNA1S Genotypes. Clinical pharmacology and therapeutics. 2019. PMID:30499100

allopurinol

DPWG

Population: Unspecified **Classification:** No recommendation

Implications: **ABCG2:rs2231142 reference (G)/rs2231142 reference (G)**
The guideline does not provide a description of the impact of the ABCG2 rs2231142 GG genotype (c.421CC; p.141QQ) on allopurinol.

Recommendation: The guideline does not provide a recommendation for allopurinol in patients with the the ABCG2 rs2231142 GG genotype (c.421CC; p.141QQ)

Citation:

- Clinical Pharmacogenetics Implementation Consortium guidelines for human leukocyte antigen-B genotype and allopurinol dosing. Clinical pharmacology and therapeutics. 2013. PMID:23232549
- Clinical Pharmacogenetics Implementation Consortium (CPIC) guidelines for human leukocyte antigen B (HLA-B) genotype and allopurinol dosing: 2015 update. Clinical pharmacology and therapeutics. 2016. PMID:26094938
- Dutch pharmacogenetics working group guideline for the gene-drug interaction of ABCG2, HLA-B and Allopurinol, and MTHFR, folic acid and methotrexate. European journal of human genetics : EJHG. 2022. PMID:36056234

Pharmacogenomics Report

John Doe (4376)

amitriptyline

CPIC

Population: General **Classification:** No recommendation

Implications: **CYP2C19:*1/*1,CYP2D6:*49/*36+*10**
CYP2C19: Normal metabolism of tertiary amines,CYP2D6: n/a

Recommendation: No recommendation

Citation:

- Clinical Pharmacogenetics Implementation Consortium guideline for CYP2D6 and CYP2C19 genotypes and dosing of tricyclic antidepressants. Clinical pharmacology and therapeutics. 2013. PMID:23486447
- Clinical pharmacogenetics implementation consortium guideline (CPIC) for CYP2D6 and CYP2C19 genotypes and dosing of tricyclic antidepressants: 2016 update. Clinical pharmacology and therapeutics. 2017. PMID:27997040
- Pharmacogenetics: from bench to byte--an update of guidelines. Clinical pharmacology and therapeutics. 2011. PMID:21412232

atomoxetine

CPIC

Population: Adults **Classification:** No recommendation

Implications: **CYP2D6:*49/*36+*10**
CYP2D6: n/a

Recommendation: No recommendation

CPIC

Population: Pediatrics **Classification:** No recommendation

Implications: **CYP2D6:*49/*36+*10**
CYP2D6: n/a

Recommendation: No recommendation

Citation:

- Clinical Pharmacogenetics Implementation Consortium Guideline for Cytochrome P450 (CYP)2D6 Genotype and Atomoxetine Therapy. Clinical pharmacology and therapeutics. 2019. PMID:30801677
- Pharmacogenetics: from bench to byte--an update of guidelines. Clinical pharmacology and therapeutics. 2011. PMID:21412232
- Dutch pharmacogenetics working group (DPWG) guideline for the gene-drug interaction of CYP2D6 and COMT with atomoxetine and methylphenidate. European journal of human genetics : EJHG. 2022. PMID:36509836

Pharmacogenomics Report

John Doe (4376)

atorvastatin

CPIC

Population: General **Classification:** Strong

Implications: **SLCO1B1:*37/*37**
SLCO1B1: Typical myopathy risk and statin exposure

Recommendation: Prescribe desired starting dose and adjust doses based on disease-specific guidelines.

Other Considerations

The potential for drug-drug interactions and dose limits based on renal and hepatic function and ancestry should be evaluated prior to initiating a statin.

Citation:

- The Clinical Pharmacogenetics Implementation Consortium Guideline for SLCO1B1, ABCG2, and CYP2C9 genotypes and Statin-Associated Musculoskeletal Symptoms. Clinical pharmacology and therapeutics. 2022. PMID:35152405

capecitabine

CPIC

Population: General **Classification:** Strong

Implications: **DPYD:Reference/c.85T>C (*9A)**
DPYD: Normal DPD activity and "normal" risk for fluoropyrimidine toxicity

Recommendation: Based on genotype, there is no indication to change dose or therapy. Use label-recommended dosage and administration.

DPWG

Population: Unspecified **Classification:** No recommendation

Implications: **DPYD:Reference/Reference**
The guideline does not provide a description of the impact of a DPYD activity score of 2 on capecitabine.

Recommendation: The guideline does not provide a recommendation for capecitabine in patients with a DPYD activity score of 2.

Citation:

- Clinical Pharmacogenetics Implementation Consortium guidelines for dihydropyrimidine dehydrogenase genotype and fluoropyrimidine dosing. Clinical pharmacology and therapeutics. 2013. PMID:23988873
- Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for Dihydropyrimidine Dehydrogenase Genotype and Fluoropyrimidine Dosing: 2017 Update. Clinical pharmacology and therapeutics. 2018. PMID:29152729
- Pharmacogenetics: from bench to byte--an update of guidelines. Clinical pharmacology and therapeutics. 2011. PMID:21412232
- Dutch Pharmacogenetics Working Group (DPWG) guideline for the gene-drug interaction of DPYD and fluoropyrimidines. European journal of human genetics : EJHG. 2019. PMID:31745289

Pharmacogenomics Report

John Doe (4376)

celecoxib

CPIC

Population: General **Classification:** Strong

Implications: **CYP2C9:*1/*1**
CYP2C9: Normal metabolism

Recommendation: Initiate therapy with recommended starting dose. In accordance with the prescribing information, use the lowest effective dosage for shortest duration consistent with individual patient treatment goals.

Citation:

- Clinical Pharmacogenetics Implementation Consortium Guideline (CPIC) for CYP2C9 and Nonsteroidal Anti-Inflammatory Drugs. Clinical pharmacology and therapeutics. 2020. PMID:32189324

citalopram

CPIC

Population: General **Classification:** Strong

Implications: **CYP2C19:*1/*1**
CYP2C19: Normal metabolism

Recommendation: Initiate therapy with recommended starting dose

DPWG

Population: Unspecified **Classification:** No recommendation

Implications: **CYP2C19:*1/*1**
The guideline does not provide a description of the impact of a normal metabolizer phenotype on citalopram.

Recommendation: The guideline does not provide a recommendation for citalopram in normal metabolizers.

Citation:

- Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for CYP2D6 and CYP2C19 Genotypes and Dosing of Selective Serotonin Reuptake Inhibitors. Clinical pharmacology and therapeutics. 2015. PMID:25974703
- Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for CYP2D6, CYP2C19, CYP2B6, SLC6A4, and HTR2A Genotypes and Serotonin Reuptake Inhibitor Antidepressants. Clinical pharmacology and therapeutics. 2023. PMID:37032427
- Pharmacogenetics: from bench to byte--an update of guidelines. Clinical pharmacology and therapeutics. 2011. PMID:21412232
- Dutch Pharmacogenetics Working Group (DPWG) guideline for the gene-drug interaction between CYP2C19 and CYP2D6 and SSRIs. European journal of human genetics : EJHG. 2022. PMID:34782755

Pharmacogenomics Report

John Doe (4376)

● clomipramine

CPIC

Population: General **Classification:** Strong

Implications: **CYP2C19:*1/*1,CYP2D6:*49/*36+*10**
CYP2C19: Normal metabolism of tertiary amines,CYP2D6: n/a

Recommendation: No recommendation

DPWG

Population: Unspecified **Classification:** No recommendation

Implications: **CYP2C19:*1/*1**
The guideline does not provide a description of the impact of a normal metabolizer phenotype on clomipramine.

Recommendation: The guideline does not provide a recommendation for clomipramine in normal metabolizers.

Citation:

- Clinical Pharmacogenetics Implementation Consortium guideline for CYP2D6 and CYP2C19 genotypes and dosing of tricyclic antidepressants. Clinical pharmacology and therapeutics. 2013. PMID:23486447
- Clinical pharmacogenetics implementation consortium guideline (CPIC) for CYP2D6 and CYP2C19 genotypes and dosing of tricyclic antidepressants: 2016 update. Clinical pharmacology and therapeutics. 2017. PMID:27997040
- Pharmacogenetics: from bench to byte--an update of guidelines. Clinical pharmacology and therapeutics. 2011. PMID:21412232

Pharmacogenomics Report

John Doe (4376)

clopidogrel

CPIC

Population: CVI ACS PCI **Classification:** Strong

Implications: **CYP2C19:*1/*1**
CYP2C19: Normal clopidogrel active metabolite formation; normal on-treatment platelet reactivity

Recommendation: If considering clopidogrel, use at standard dose (75 mg/day)

Other Considerations

For cardiovascular indications of acute coronary syndrome (ACS) and/or percutaneous coronary intervention (PCI). ACS and/or PCI includes patients undergoing PCI for an ACS or non-ACS (elective) indication.

CPIC

Population: CVI Non-ACS Non-PCI **Classification:** Strong

Implications: **CYP2C19:*1/*1**
CYP2C19: Normal clopidogrel active metabolite formation; normal on-treatment platelet reactivity

Recommendation: If considering clopidogrel, use at standard dose (75 mg/day)

Other Considerations

For non-acute coronary syndrome (non-ACS) and non-percutaneous coronary intervention (non-PCI) cardiovascular indications. Non-ACS, non-PCI cardiovascular indications include peripheral arterial disease and stable coronary artery disease following a recent myocardial infarction outside the setting of PCI.

CPIC

Population: NVI **Classification:** Strong

Implications: **CYP2C19:*1/*1**
CYP2C19: Normal clopidogrel active metabolite formation; normal on-treatment platelet reactivity

Recommendation: If considering clopidogrel, use at standard dose (75 mg/day)

Other Considerations

For neurovascular indications. Neurovascular disease includes acute ischemic stroke or transient ischemic attack, secondary prevention of stroke, or prevention of thromboembolic events following neurointerventional procedures such as carotid artery stenting and stent-assisted coiling of intracranial aneurysms.

DPWG

Population: Unspecified **Classification:** No recommendation

Implications: **CYP2C19:*1/*1**
The guideline does not provide a description of the impact of a normal metabolizer phenotype on clopidogrel.

Recommendation: The guideline does not provide a recommendation for clopidogrel in normal metabolizers.

Citation:

- Clinical Pharmacogenetics Implementation Consortium guidelines for cytochrome P450-2C19 (CYP2C19) genotype and clopidogrel therapy. Clinical pharmacology and therapeutics. 2011. PMID:21716271
- Clinical Pharmacogenetics Implementation Consortium guidelines for CYP2C19 genotype and clopidogrel therapy: 2013 update. Clinical pharmacology and therapeutics. 2013. PMID:23698643
- Clinical Pharmacogenetics Implementation Consortium Guideline for CYP2C19 Genotype and Clopidogrel Therapy: 2022 Update. Clinical pharmacology and therapeutics. 2022. PMID:35034351
- Pharmacogenetics: from bench to byte--an update of guidelines. Clinical pharmacology and therapeutics. 2011. PMID:21412232

codeine

CPIC

Population: General **Classification:** No recommendation

Implications: **CYP2D6:*49/*36+*10**
CYP2D6: n/a

Recommendation: No recommendation

Citation:

- Clinical Pharmacogenetics Implementation Consortium (CPIC) guidelines for codeine therapy in the context of cytochrome P450 2D6 (CYP2D6) genotype. Clinical pharmacology and therapeutics. 2012. PMID:22205192
- Clinical Pharmacogenetics Implementation Consortium guidelines for cytochrome P450 2D6 genotype and codeine therapy: 2014 update. Clinical pharmacology and therapeutics. 2014. PMID:24458010
- Clinical Pharmacogenetics Implementation Consortium Guideline for CYP2D6, OPRM1, and COMT Genotypes and Select Opioid Therapy. Clinical pharmacology and therapeutics. 2021. PMID:33387367
- Pharmacogenetics: from bench to byte--an update of guidelines. Clinical pharmacology and therapeutics. 2011. PMID:21412232
- Dutch Pharmacogenetics Working Group (DPWG) guideline for the gene-drug interaction between CYP2D6 and opioids (codeine, tramadol and oxycodone). European journal of human genetics : EJHG. 2022. PMID:34267337

dapsone

CPIC

Population: General **Classification:** Strong

Implications: **G6PD:B (reference)/B (reference)**
G6PD: Low risk of acute hemolytic anemia

Recommendation: No reason to avoid based on G6PD status

Citation:

- Clinical Pharmacogenetics Implementation Consortium (CPIC) guidelines for rasburicase therapy in the context of G6PD deficiency genotype. Clinical pharmacology and therapeutics. 2014. PMID:24787449
- Expanded Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for Medication Use in the Context of G6PD Genotype. Clinical pharmacology and therapeutics. 2022. PMID:36049896

Pharmacogenomics Report

John Doe (4376)

desipramine

CPIC

Population: General **Classification:** No recommendation

Implications: **CYP2D6:*49/*36+*10**
CYP2D6: n/a

Recommendation: No recommendation

Citation:

- Clinical Pharmacogenetics Implementation Consortium guideline for CYP2D6 and CYP2C19 genotypes and dosing of tricyclic antidepressants. Clinical pharmacology and therapeutics. 2013. PMID:23486447
- Clinical pharmacogenetics implementation consortium guideline (CPIC) for CYP2D6 and CYP2C19 genotypes and dosing of tricyclic antidepressants: 2016 update. Clinical pharmacology and therapeutics. 2017. PMID:27997040

doxepin

CPIC

Population: General **Classification:** Strong

Implications: **CYP2C19:*1/*1,CYP2D6:*49/*36+*10**
CYP2C19: Normal metabolism of tertiary amines,CYP2D6: n/a

Recommendation: No recommendation

Citation:

- Clinical Pharmacogenetics Implementation Consortium guideline for CYP2D6 and CYP2C19 genotypes and dosing of tricyclic antidepressants. Clinical pharmacology and therapeutics. 2013. PMID:23486447
- Clinical pharmacogenetics implementation consortium guideline (CPIC) for CYP2D6 and CYP2C19 genotypes and dosing of tricyclic antidepressants: 2016 update. Clinical pharmacology and therapeutics. 2017. PMID:27997040
- Pharmacogenetics: from bench to byte--an update of guidelines. Clinical pharmacology and therapeutics. 2011. PMID:21412232

Pharmacogenomics Report

John Doe (4376)

efavirenz

CPIC

Population: Child >40kg_adult **Classification:** Strong

Implications: **CYP2B6:*1/*1**
CYP2B6: Normal efavirenz metabolism

Recommendation: Initiate efavirenz with standard dosing (600 mg/day)

Other Considerations

The ENCORE study showed that in treatment-naïve patients randomized to initiate efavirenz-based regimens (combined with tenofovir and emtricitabine), 400 mg/day was non-inferior to 600 mg/day regardless of CYP2B6 genotype (PMID 24522178).

DPWG

Population: Unspecified **Classification:** No recommendation

Implications: **CYP2B6:*1/*1**
The guideline does not provide a description of the impact of a normal metabolizer phenotype on efavirenz.

Recommendation: The guideline does not provide a recommendation for efavirenz in normal metabolizers.

Citation:

- Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for CYP2B6 and Efavirenz-Containing Antiretroviral Therapy. Clinical pharmacology and therapeutics. 2019. PMID:31006110

escitalopram

CPIC

Population: General **Classification:** Strong

Implications: **CYP2C19:*1/*1**
CYP2C19: Normal metabolism

Recommendation: Initiate therapy with recommended starting dose

DPWG

Population: Unspecified **Classification:** No recommendation

Implications: **CYP2C19:*1/*1**
The guideline does not provide a description of the impact of a normal metabolizer phenotype on escitalopram.

Recommendation: The guideline does not provide a recommendation for escitalopram in normal metabolizers.

Citation:

- Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for CYP2D6 and CYP2C19 Genotypes and Dosing of Selective Serotonin Reuptake Inhibitors. Clinical pharmacology and therapeutics. 2015. PMID:25974703
- Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for CYP2D6, CYP2C19, CYP2B6, SLC6A4, and HTR2A Genotypes and Serotonin Reuptake Inhibitor Antidepressants. Clinical pharmacology and therapeutics. 2023. PMID:37032427
- Pharmacogenetics: from bench to byte--an update of guidelines. Clinical pharmacology and therapeutics. 2011. PMID:21412232
- Dutch Pharmacogenetics Working Group (DPWG) guideline for the gene-drug interaction between CYP2C19 and CYP2D6 and SSRIs. European journal of human genetics : EJHG. 2022. PMID:34782755

Pharmacogenomics Report

John Doe (4376)

● flucytosine

DPWG

Population: Unspecified **Classification:** No recommendation

Implications: **DPYD:Reference/Reference**
The guideline does not provide a description of the impact of a DPYD activity score of 2 on flucytosine.

Recommendation: The guideline does not provide a recommendation for flucytosine in patients with a DPYD activity score of 2.

Citation: N/A

● fluorouracil

CPIC

Population: General **Classification:** Strong

Implications: **DPYD:Reference/c.85T>C (*9A)**
DPYD: Normal DPD activity and "normal" risk for fluoropyrimidine toxicity

Recommendation: Based on genotype, there is no indication to change dose or therapy. Use label-recommended dosage and administration.

DPWG

Population: Unspecified **Classification:** No recommendation

Implications: **DPYD:Reference/Reference**
The guideline does not provide a description of the impact of a DPYD activity score of 2 on fluorouracil.

Recommendation: The guideline does not provide a recommendation for fluorouracil in patients with a DPYD activity score of 2.

Citation:

- Clinical Pharmacogenetics Implementation Consortium guidelines for dihydropyrimidine dehydrogenase genotype and fluoropyrimidine dosing. Clinical pharmacology and therapeutics. 2013. PMID:23988873
- Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for Dihydropyrimidine Dehydrogenase Genotype and Fluoropyrimidine Dosing: 2017 Update. Clinical pharmacology and therapeutics. 2018. PMID:29152729
- Pharmacogenetics: from bench to byte--an update of guidelines. Clinical pharmacology and therapeutics. 2011. PMID:21412232
- Dutch Pharmacogenetics Working Group (DPWG) guideline for the gene-drug interaction of DPYD and fluoropyrimidines. European journal of human genetics : EJHG. 2019. PMID:31745289

Pharmacogenomics Report

John Doe (4376)

flurbiprofen

CPIC

Population: General **Classification:** Strong

Implications: **CYP2C9:*1/*1**
CYP2C9: Normal metabolism

Recommendation: Initiate therapy with recommended starting dose. In accordance with the prescribing information, use the lowest effective dosage for shortest duration consistent with individual patient treatment goals.

Citation:

- Clinical Pharmacogenetics Implementation Consortium Guideline (CPIC) for CYP2C9 and Nonsteroidal Anti-Inflammatory Drugs. Clinical pharmacology and therapeutics. 2020. PMID:32189324

fluvastatin

CPIC

Population: General **Classification:** Strong

Implications: **CYP2C9:*1/*1,SLCO1B1:*37/*37**
CYP2C9: Normal exposure.,SLCO1B1: Typical myopathy risk and statin exposure.

Recommendation: Prescribe desired starting dose and adjust doses of fluvastatin based on disease-specific guidelines.

Other Considerations

The potential for drug-drug interactions and dose limits based on renal and hepatic function and ancestry should be evaluated prior to initiating a statin.

Citation:

- The Clinical Pharmacogenetics Implementation Consortium Guideline for SLCO1B1, ABCG2, and CYP2C9 genotypes and Statin-Associated Musculoskeletal Symptoms. Clinical pharmacology and therapeutics. 2022. PMID:35152405

fluvoxamine

CPIC

Population: General **Classification:** No recommendation

Implications: **CYP2D6:*49/*36+*10**
CYP2D6: n/a

Recommendation: No recommendation

Citation:

- Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for CYP2D6 and CYP2C19 Genotypes and Dosing of Selective Serotonin Reuptake Inhibitors. Clinical pharmacology and therapeutics. 2015. PMID:25974703
- Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for CYP2D6, CYP2C19, CYP2B6, SLC6A4, and HTR2A Genotypes and Serotonin Reuptake Inhibitor Antidepressants. Clinical pharmacology and therapeutics. 2023. PMID:37032427

fosphenytoin

CPIC

Population: PHT Naive **Classification:** Strong

Implications: **CYP2C9:*1/*1,HLA-B:Unknown/Unknown**
CYP2C9: Normal phenytoin metabolism,HLA-B: n/a

Recommendation: No adjustments needed from typical dosing strategies. Subsequent doses should be adjusted according to therapeutic drug monitoring, response, and side effects. An HLA-B*15:02 negative test does not eliminate the risk of phenytoin-induced SJS/TEN and patients should be carefully monitored according to a usual standard.

CPIC

Population: PHT Use >3mos **Classification:** Strong

Implications: **CYP2C9:*1/*1,HLA-B:Unknown/Unknown**
CYP2C9: Normal phenytoin metabolism,HLA-B: n/a

Recommendation: No adjustments needed from typical dosing strategies. Subsequent doses should be adjusted according to therapeutic drug monitoring, response, and side effects. An HLA-B*15:02 negative test does not eliminate the risk of phenytoin-induced SJS/TEN and patients should be carefully monitored according to a usual standard.

Citation:

- Clinical pharmacogenetics implementation consortium guidelines for CYP2C9 and HLA-B genotypes and phenytoin dosing. Clinical pharmacology and therapeutics. 2014. PMID:25099164
- Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for CYP2C9 and HLA-B Genotypes and Phenytoin Dosing: 2020 Update. Clinical pharmacology and therapeutics. 2021. PMID:32779747

hormonal contraceptives for systemic use

DPWG

Population: Unspecified **Classification:** No recommendation

Implications: **F5:rs6025 C/rs6025 C**
The guideline does not provide a description of the impact of the absence of Factor V Leiden on estrogen-containing contraceptives.

Recommendation: The guideline does not provide a recommendation for estrogen-containing contraceptives in patients who do not have the Factor V Leiden variant.

Citation:

- Pharmacogenetics: from bench to byte--an update of guidelines. Clinical pharmacology and therapeutics. 2011. PMID:21412232

Pharmacogenomics Report

John Doe (4376)

hydrocodone

CPIC

Population: General **Classification:** No recommendation

Implications: **CYP2D6:*49/*36+*10**
CYP2D6: n/a

Recommendation: No recommendation

Citation:

- Clinical Pharmacogenetics Implementation Consortium Guideline for CYP2D6, OPRM1, and COMT Genotypes and Select Opioid Therapy. Clinical pharmacology and therapeutics. 2021. PMID:33387367

ibuprofen

CPIC

Population: General **Classification:** Strong

Implications: **CYP2C9:*1/*1**
CYP2C9: Normal metabolism

Recommendation: Initiate therapy with recommended starting dose. In accordance with the prescribing information, use the lowest effective dosage for shortest duration consistent with individual patient treatment goals.

Citation:

- Clinical Pharmacogenetics Implementation Consortium Guideline (CPIC) for CYP2C9 and Nonsteroidal Anti-Inflammatory Drugs. Clinical pharmacology and therapeutics. 2020. PMID:32189324

Pharmacogenomics Report

John Doe (4376)

imipramine

CPIC

Population: General **Classification:** Strong

Implications: **CYP2C19:*1/*1,CYP2D6:*49/*36+*10**
CYP2C19: Normal metabolism of tertiary amines,CYP2D6: n/a

Recommendation: No recommendation

DPWG

Population: Unspecified **Classification:** No recommendation

Implications: **CYP2C19:*1/*1**
The guideline does not provide a description of the impact of a normal metabolizer phenotype on imipramine.

Recommendation: The guideline does not provide a recommendation for imipramine in normal metabolizers.

Citation:

- Clinical Pharmacogenetics Implementation Consortium guideline for CYP2D6 and CYP2C19 genotypes and dosing of tricyclic antidepressants. Clinical pharmacology and therapeutics. 2013. PMID:23486447
- Clinical pharmacogenetics implementation consortium guideline (CPIC) for CYP2D6 and CYP2C19 genotypes and dosing of tricyclic antidepressants: 2016 update. Clinical pharmacology and therapeutics. 2017. PMID:27997040
- Pharmacogenetics: from bench to byte--an update of guidelines. Clinical pharmacology and therapeutics. 2011. PMID:21412232

irinotecan

DPWG

Population: Unspecified **Classification:** N/A

Implications: **UGT1A1:*1/*6**
This genetic variation (IM) is more common in Western populations than the wild-type (*1/*1). This means that treatment is largely geared to patients with this genetic variation. Adjustment of the treatment is therefore not useful.

Recommendation: NO action is needed for this gene-drug interaction.

Citation:

- Pharmacogenetics: from bench to byte--an update of guidelines. Clinical pharmacology and therapeutics. 2011. PMID:21412232
- Dutch pharmacogenetics working group (DPWG) guideline for the gene-drug interaction between UGT1A1 and irinotecan. European journal of human genetics : EJHG. 2023. PMID:36443464

Pharmacogenomics Report

John Doe (4376)

● lornoxicam

CPIC

Population: General **Classification:** Strong

Implications:
CYP2C9:*1/*1
CYP2C9: Normal metabolism

Recommendation: Initiate therapy with recommended starting dose. In accordance with the prescribing information, use the lowest effective dosage for shortest duration consistent with individual patient treatment goals.

Citation:
- Clinical Pharmacogenetics Implementation Consortium Guideline (CPIC) for CYP2C9 and Nonsteroidal Anti-Inflammatory Drugs. Clinical pharmacology and therapeutics. 2020. PMID:32189324

● lovastatin

CPIC

Population: General **Classification:** Strong

Implications:
SLCO1B1:*37/*37
SLCO1B1: Typical myopathy risk and statin exposure

Recommendation: Prescribe desired starting dose and adjust doses based on disease-specific guidelines.

Other Considerations

The potential for drug-drug interactions and dose limits based on renal and hepatic function and ancestry should be evaluated prior to initiating a statin.

Citation:
- The Clinical Pharmacogenetics Implementation Consortium Guideline for SLCO1B1, ABCG2, and CYP2C9 genotypes and Statin-Associated Musculoskeletal Symptoms. Clinical pharmacology and therapeutics. 2022. PMID:35152405

● meloxicam

CPIC

Population: General **Classification:** Strong

Implications:
CYP2C9:*1/*1
CYP2C9: Normal metabolism

Recommendation: Initiate therapy with recommended starting dose. In accordance with the prescribing information, use the lowest effective dosage for shortest duration consistent with individual patient treatment goals.

Citation:
- Clinical Pharmacogenetics Implementation Consortium Guideline (CPIC) for CYP2C9 and Nonsteroidal Anti-Inflammatory Drugs. Clinical pharmacology and therapeutics. 2020. PMID:32189324

Pharmacogenomics Report

John Doe (4376)

methylene blue

CPIC

Population: General **Classification:** Strong

Implications: **G6PD:B (reference)/B (reference)**
G6PD: Low risk of acute hemolytic anemia

Recommendation: No reason to avoid based on G6PD status

Citation:

- Clinical Pharmacogenetics Implementation Consortium (CPIC) guidelines for rasburicase therapy in the context of G6PD deficiency genotype. Clinical pharmacology and therapeutics. 2014. PMID:24787449
- Expanded Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for Medication Use in the Context of G6PD Genotype. Clinical pharmacology and therapeutics. 2022. PMID:36049896

nitrofurantoin

CPIC

Population: General **Classification:** Strong

Implications: **G6PD:B (reference)/B (reference)**
G6PD: Low risk of acute hemolytic anemia

Recommendation: No reason to avoid based on G6PD status

Citation: N/A

nortriptyline

CPIC

Population: General **Classification:** No recommendation

Implications: **CYP2D6:*49/*36+*10**
CYP2D6: n/a

Recommendation: No recommendation

Other Considerations

Because CYP2D6 phenotype could not be assigned based on genotyping performed, therapuetic monitoring should be considered. If therapuetic monitoring cannot be performed, monitor closely for toxicity and/or efficacy.

Citation:

- Clinical Pharmacogenetics Implementation Consortium guideline for CYP2D6 and CYP2C19 genotypes and dosing of tricyclic antidepressants. Clinical pharmacology and therapeutics. 2013. PMID:23486447
- Clinical pharmacogenetics implementation consortium guideline (CPIC) for CYP2D6 and CYP2C19 genotypes and dosing of tricyclic antidepressants: 2016 update. Clinical pharmacology and therapeutics. 2017. PMID:27997040
- Pharmacogenetics: from bench to byte--an update of guidelines. Clinical pharmacology and therapeutics. 2011. PMID:21412232

Pharmacogenomics Report

John Doe (4376)

● ondansetron

CPIC

Population: General **Classification:** No recommendation

Implications: **CYP2D6:*49/*36+*10**
CYP2D6: n/a

Recommendation: No recommendation

Citation:

- Clinical Pharmacogenetics Implementation Consortium (CPIC) guideline for CYP2D6 genotype and use of ondansetron and tropisetron. Clinical pharmacology and therapeutics. 2017. PMID:28002639

● paroxetine

CPIC

Population: General **Classification:** No recommendation

Implications: **CYP2D6:*49/*36+*10**
CYP2D6: n/a

Recommendation: No recommendation

Citation:

- Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for CYP2D6 and CYP2C19 Genotypes and Dosing of Selective Serotonin Reuptake Inhibitors. Clinical pharmacology and therapeutics. 2015. PMID:25974703
- Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for CYP2D6, CYP2C19, CYP2B6, SLC6A4, and HTR2A Genotypes and Serotonin Reuptake Inhibitor Antidepressants. Clinical pharmacology and therapeutics. 2023. PMID:37032427
- Pharmacogenetics: from bench to byte--an update of guidelines. Clinical pharmacology and therapeutics. 2011. PMID:21412232
- Dutch Pharmacogenetics Working Group (DPWG) guideline for the gene-drug interaction between CYP2C19 and CYP2D6 and SSRIs. European journal of human genetics : EJHG. 2022. PMID:34782755

● pegloticase

CPIC

Population: General **Classification:** Strong

Implications: **G6PD:B (reference)/B (reference)**
G6PD: Low risk of acute hemolytic anemia

Recommendation: No reason to avoid based on G6PD status

Citation:

- Clinical Pharmacogenetics Implementation Consortium (CPIC) guidelines for rasburicase therapy in the context of G6PD deficiency genotype. Clinical pharmacology and therapeutics. 2014. PMID:24787449
- Expanded Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for Medication Use in the Context of G6PD Genotype. Clinical pharmacology and therapeutics. 2022. PMID:36049896

phenytoin

CPIC

Population: PHT Naive **Classification:** Strong

Implications: **CYP2C9:*1/*1,HLA-B:Unknown/Unknown**
CYP2C9: Normal phenytoin metabolism,HLA-B: n/a

Recommendation: No adjustments needed from typical dosing strategies. Subsequent doses should be adjusted according to therapeutic drug monitoring, response, and side effects. An HLA-B*15:02 negative test does not eliminate the risk of phenytoin-induced SJS/TEN and patients should be carefully monitored according to a usual standard.

CPIC

Population: PHT Use >3mos **Classification:** Strong

Implications: **CYP2C9:*1/*1,HLA-B:Unknown/Unknown**
CYP2C9: Normal phenytoin metabolism,HLA-B: n/a

Recommendation: No adjustments needed from typical dosing strategies. Subsequent doses should be adjusted according to therapeutic drug monitoring, response, and side effects. An HLA-B*15:02 negative test does not eliminate the risk of phenytoin-induced SJS/TEN and patients should be carefully monitored according to a usual standard.

DPWG

Population: Unspecified **Classification:** No recommendation

Implications: **CYP2C9:*1/*1**
The guideline does not provide a description of the impact of a normal metabolizer phenotype on phenytoin.

Recommendation: The guideline does not provide a recommendation for phenytoin in normal metabolizers.

Citation:

- Clinical pharmacogenetics implementation consortium guidelines for CYP2C9 and HLA-B genotypes and phenytoin dosing. Clinical pharmacology and therapeutics. 2014. PMID:25099164
- Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for CYP2C9 and HLA-B Genotypes and Phenytoin Dosing: 2020 Update. Clinical pharmacology and therapeutics. 2021. PMID:32779747
- Pharmacogenetics: from bench to byte--an update of guidelines. Clinical pharmacology and therapeutics. 2011. PMID:21412232

piroxicam

CPIC

Population: General **Classification:** Strong

Implications: **CYP2C9:*1/*1**
CYP2C9: Normal metabolism

Recommendation: Initiate therapy with recommended starting dose. In accordance with the prescribing information, use the lowest effective dosage for shortest duration consistent with individual patient treatment goals.

Citation:

- Clinical Pharmacogenetics Implementation Consortium Guideline (CPIC) for CYP2C9 and Nonsteroidal Anti-Inflammatory Drugs. Clinical pharmacology and therapeutics. 2020. PMID:32189324

Pharmacogenomics Report

John Doe (4376)

pitavastatin

CPIC

Population: General **Classification:** Strong

Implications: **SLCO1B1:*37/*37**
SLCO1B1: Typical myopathy risk and statin exposure

Recommendation: Prescribe desired starting dose and adjust doses based on disease-specific guidelines.

Other Considerations

The potential for drug-drug interactions and dose limits based on renal and hepatic function and ancestry should be evaluated prior to initiating a statin.

Citation:

- The Clinical Pharmacogenetics Implementation Consortium Guideline for SLCO1B1, ABCG2, and CYP2C9 genotypes and Statin-Associated Musculoskeletal Symptoms. Clinical pharmacology and therapeutics. 2022. PMID:35152405

pravastatin

CPIC

Population: General **Classification:** Strong

Implications: **SLCO1B1:*37/*37**
SLCO1B1: Typical myopathy risk and statin exposure

Recommendation: Prescribe desired starting dose and adjust doses based on disease-specific guidelines.

Other Considerations

The potential for drug-drug interactions and dose limits based on renal and hepatic function and ancestry should be evaluated prior to initiating a statin.

Citation:

- The Clinical Pharmacogenetics Implementation Consortium Guideline for SLCO1B1, ABCG2, and CYP2C9 genotypes and Statin-Associated Musculoskeletal Symptoms. Clinical pharmacology and therapeutics. 2022. PMID:35152405

primaquine

CPIC

Population: General **Classification:** Strong

Implications: **G6PD:B (reference)/B (reference)**
G6PD: Low risk of acute hemolytic anemia

Recommendation: No reason to avoid based on G6PD status

Citation:

- Expanded Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for Medication Use in the Context of G6PD Genotype. Clinical pharmacology and therapeutics. 2022. PMID:36049896

Pharmacogenomics Report

John Doe (4376)

quetiapine

DPWG

Population: Unspecified **Classification:** No recommendation

Implications: **CYP3A4:*1/*1**
The guideline does not provide a description of the impact of a normal metabolizer phenotype on quetiapine.

Recommendation: The guideline does not provide a recommendation for quetiapine in normal metabolizers.

Citation:

- Dutch Pharmacogenetics Working Group (DPWG) guideline for the gene-drug interaction between CYP2D6, CYP3A4 and CYP1A2 and antipsychotics. European journal of human genetics : EJHG. 2023. PMID:37002327

rasburicase

CPIC

Population: General **Classification:** Strong

Implications: **G6PD:B (reference)/B (reference)**
G6PD: Low risk of acute hemolytic anemia

Recommendation: No reason to avoid based on G6PD status

Citation:

- Clinical Pharmacogenetics Implementation Consortium (CPIC) guidelines for rasburicase therapy in the context of G6PD deficiency genotype. Clinical pharmacology and therapeutics. 2014. PMID:24787449
- Expanded Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for Medication Use in the Context of G6PD Genotype. Clinical pharmacology and therapeutics. 2022. PMID:36049896

rosuvastatin

CPIC

Population: General **Classification:** Strong

Implications: **ABCG2:rs2231142 reference (G)/rs2231142 reference (G),SLCO1B1:*37/*37**
ABCG2: Typical myopathy risk and rosuvastatin exposure,SLCO1B1: Typical myopathy risk and statin exposure

Recommendation: Prescribe desired starting dose and adjust doses of rosuvastatin based on disease-specific and specific population guidelines.

Other Considerations

The potential for drug-drug interactions and dose limits based on renal and hepatic function and Asian ancestry should be evaluated prior to initiating rosuvastatin.

Citation:

- The Clinical Pharmacogenetics Implementation Consortium Guideline for SLCO1B1, ABCG2, and CYP2C9 genotypes and Statin-Associated Musculoskeletal Symptoms. Clinical pharmacology and therapeutics. 2022. PMID:35152405

Pharmacogenomics Report

John Doe (4376)

● sertraline

CPIC

Population: General **Classification:** Strong

Implications: **CYP2B6:*1/*1,CYP2C19:*1/*1**
CYP2B6: Normal metabolism of sertraline to less active compounds.,CYP2C19: Normal metabolism

Recommendation: Initiate therapy with recommended starting dose.

DPWG

Population: Unspecified **Classification:** No recommendation

Implications: **CYP2C19:*1/*1**
The guideline does not provide a description of the impact of a normal metabolizer phenotype on sertraline.

Recommendation: The guideline does not provide a recommendation for sertraline in normal metabolizers.

Citation:

- Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for CYP2D6 and CYP2C19 Genotypes and Dosing of Selective Serotonin Reuptake Inhibitors. Clinical pharmacology and therapeutics. 2015. PMID:25974703
- Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for CYP2D6, CYP2C19, CYP2B6, SLC6A4, and HTR2A Genotypes and Serotonin Reuptake Inhibitor Antidepressants. Clinical pharmacology and therapeutics. 2023. PMID:37032427
- Pharmacogenetics: from bench to byte--an update of guidelines. Clinical pharmacology and therapeutics. 2011. PMID:21412232
- Dutch Pharmacogenetics Working Group (DPWG) guideline for the gene-drug interaction between CYP2C19 and CYP2D6 and SSRIs. European journal of human genetics : EJHG. 2022. PMID:34782755

● simvastatin

CPIC

Population: General **Classification:** Strong

Implications: **SLCO1B1:*37/*37**
SLCO1B1: Typical myopathy risk and statin exposure

Recommendation: Prescribe desired starting dose and adjust doses based on disease-specific guidelines.

Other Considerations
The potential for drug-drug interactions and dose limits based on renal and hepatic function and ancestry should be evaluated prior to initiating a statin.

Citation:

- The clinical pharmacogenomics implementation consortium: CPIC guideline for SLCO1B1 and simvastatin-induced myopathy. Clinical pharmacology and therapeutics. 2012. PMID:22617227
- The clinical pharmacogenetics implementation consortium guideline for SLCO1B1 and simvastatin-induced myopathy: 2014 update. Clinical pharmacology and therapeutics. 2014. PMID:24918167
- The Clinical Pharmacogenetics Implementation Consortium Guideline for SLCO1B1, ABCG2, and CYP2C9 genotypes and Statin-Associated Musculoskeletal Symptoms. Clinical pharmacology and therapeutics. 2022. PMID:35152405

Pharmacogenomics Report

John Doe (4376)

siponimod

DPWG

Population: Unspecified **Classification:** No recommendation

Implications: **CYP2C9:*1/*1**
The guideline does not provide a description of the impact of a normal metabolizer phenotype on siponimod.

Recommendation: The guideline does not provide a recommendation for siponimod in normal metabolizers.

Citation: N/A

tacrolimus

CPIC

Population: General **Classification:** Strong

Implications: **CYP3A5:*3/*3**
CYP3A5: Higher ("normal") dose-adjusted trough concentrations of tacrolimus and increased chance of achieving target tacrolimus concentrations.

Recommendation: Initiate therapy with standard recommended dose. Use therapeutic drug monitoring to guide dose adjustments.

Other Considerations

This recommendation includes the use of tacrolimus in kidney, heart, lung and hematopoietic stem cell transplant patients, and liver transplant patients where the donor and recipient genotypes are identical. Typically with other CYP enzymes, a normal metabolizer would be classified as having normal metabolism, and therefore, the drug dose would not change based on the patient's genotype. However, in the case of CYP3A5 and tacrolimus, a CYP3A5 expresser (i.e., CYP3A5 normal metabolizer or intermediate metabolizer) would require a higher recommended starting dose and the CYP3A5 non-expresser (i.e., poor metabolizer) would require the standard recommended starting dose.

DPWG

Population: Unspecified **Classification:** No recommendation

Implications: **CYP3A5:*3/*3**
The guideline does not provide a description of the impact of a non-expressor phenotype on tacrolimus.

Recommendation: The guideline does not provide a recommendation for tacrolimus in CYP3A5 non-expressers.

Citation:

- Clinical Pharmacogenetics Implementation Consortium (CPIC) Guidelines for CYP3A5 Genotype and Tacrolimus Dosing. Clinical pharmacology and therapeutics. 2015. PMID:25801146
- Pharmacogenetics: from bench to byte--an update of guidelines. Clinical pharmacology and therapeutics. 2011. PMID:21412232

Pharmacogenomics Report

John Doe (4376)

tafenoquine

CPIC

Population: General **Classification:** Strong

Implications: **G6PD:B (reference)/B (reference)**
G6PD: Low risk of acute hemolytic anemia

Recommendation: No reason to avoid based on G6PD status

Other Considerations

Tafenoquine's safety has been established for a G6PD enzyme activity ≥70% of normal. (Inclusion criteria for clinical trials involving tafenoquine included G6PD activity ≥70%.)

Citation:

- Clinical Pharmacogenetics Implementation Consortium (CPIC) guidelines for rasburicase therapy in the context of G6PD deficiency genotype. Clinical pharmacology and therapeutics. 2014. PMID:24787449
- Expanded Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for Medication Use in the Context of G6PD Genotype. Clinical pharmacology and therapeutics. 2022. PMID:36049896

tamoxifen

CPIC

Population: General **Classification:** No recommendation

Implications: **CYP2D6:*49/*36+*10**
CYP2D6: n/a

Recommendation: No recommendation

Citation:

- Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for CYP2D6 and Tamoxifen Therapy. Clinical pharmacology and therapeutics. 2018. PMID:29385237
- Pharmacogenetics: from bench to byte--an update of guidelines. Clinical pharmacology and therapeutics. 2011. PMID:21412232

tegafur

DPWG

Population: Unspecified **Classification:** No recommendation

Implications: **DPYD:Reference/Reference**
The guideline does not provide a description of the impact of a DPYD activity score of 2 on tegafur.

Recommendation: The guideline does not provide a recommendation for tegafur in patients with a DPYD activity score of 2.

Citation:

- Pharmacogenetics: from bench to byte--an update of guidelines. Clinical pharmacology and therapeutics. 2011. PMID:21412232
- Dutch Pharmacogenetics Working Group (DPWG) guideline for the gene-drug interaction of DPYD and fluoropyrimidines. European journal of human genetics : EJHG. 2019. PMID:31745289

tenoxicam

CPIC

Population: General **Classification:** Strong

Implications: **CYP2C9:*1/*1**
CYP2C9: Normal metabolism

Recommendation: Initiate therapy with recommended starting dose. In accordance with the prescribing information, use the lowest effective dosage for shortest duration consistent with individual patient treatment goals.

Citation:

- Clinical Pharmacogenetics Implementation Consortium Guideline (CPIC) for CYP2C9 and Nonsteroidal Anti-Inflammatory Drugs. Clinical pharmacology and therapeutics. 2020. PMID:32189324

toluidine blue

CPIC

Population: General **Classification:** Strong

Implications: **G6PD:B (reference)/B (reference)**
G6PD: Low risk of acute hemolytic anemia

Recommendation: No reason to avoid based on G6PD status

Other Considerations

Toluidine blue classification strength is based on extrapolation from methylene blue data

Citation:

- Clinical Pharmacogenetics Implementation Consortium (CPIC) guidelines for rasburicase therapy in the context of G6PD deficiency genotype. Clinical pharmacology and therapeutics. 2014. PMID:24787449
- Expanded Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for Medication Use in the Context of G6PD Genotype. Clinical pharmacology and therapeutics. 2022. PMID:36049896

tramadol

CPIC

Population: General **Classification:** No recommendation

Implications: **CYP2D6:*49/*36+*10**
CYP2D6: n/a

Recommendation: No recommendation

Citation:

- Clinical Pharmacogenetics Implementation Consortium Guideline for CYP2D6, OPRM1, and COMT Genotypes and Select Opioid Therapy. Clinical pharmacology and therapeutics. 2021. PMID:33387367
- Pharmacogenetics: from bench to byte--an update of guidelines. Clinical pharmacology and therapeutics. 2011. PMID:21412232
- Dutch Pharmacogenetics Working Group (DPWG) guideline for the gene-drug interaction between CYP2D6 and opioids (codeine, tramadol and oxycodone). European journal of human genetics : EJHG. 2022. PMID:34267337

Pharmacogenomics Report

John Doe (4376)

trimipramine

CPIC

Population: General **Classification:** Strong

Implications: **CYP2C19:*1/*1,CYP2D6:*49/*36+*10**
CYP2C19: Normal metabolism of tertiary amines,CYP2D6: n/a

Recommendation: No recommendation

Citation:

- Clinical Pharmacogenetics Implementation Consortium guideline for CYP2D6 and CYP2C19 genotypes and dosing of tricyclic antidepressants. Clinical pharmacology and therapeutics. 2013. PMID:23486447
- Clinical pharmacogenetics implementation consortium guideline (CPIC) for CYP2D6 and CYP2C19 genotypes and dosing of tricyclic antidepressants: 2016 update. Clinical pharmacology and therapeutics. 2017. PMID:27997040

tropisetron

CPIC

Population: General **Classification:** No recommendation

Implications: **CYP2D6:*49/*36+*10**
CYP2D6: n/a

Recommendation: No recommendation

Citation:

- Clinical Pharmacogenetics Implementation Consortium (CPIC) guideline for CYP2D6 genotype and use of ondansetron and tropisetron. Clinical pharmacology and therapeutics. 2017. PMID:28002639

venlafaxine

CPIC

Population: General **Classification:** No recommendation

Implications: **CYP2D6:*49/*36+*10**
CYP2D6: n/a

Recommendation: No recommendation

Citation:

- Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for CYP2D6, CYP2C19, CYP2B6, SLC6A4, and HTR2A Genotypes and Serotonin Reuptake Inhibitor Antidepressants. Clinical pharmacology and therapeutics. 2023. PMID:37032427
- Pharmacogenetics: from bench to byte--an update of guidelines. Clinical pharmacology and therapeutics. 2011. PMID:21412232

voriconazole

CPIC

Population: Adults **Classification:** Strong

Implications: **CYP2C19:*1/*1**
CYP2C19: Normal voriconazole metabolism

Recommendation: Initiate therapy with recommended standard of care dosing

Other Considerations

Further dose adjustments or selection of alternative therapy may be necessary due to other clinical factors, such as drug interactions, hepatic function, renal function, species, site of infection, therapeutic drug monitoring, and comorbidities.

CPIC

Population: Pediatrics **Classification:** Strong

Implications: **CYP2C19:*1/*1**
CYP2C19: Normal voriconazole metabolism

Recommendation: Initiate therapy with recommended standard of care dosing

Other Considerations

Further dose adjustments or selection of alternative therapy may be necessary due to other clinical factors, such as drug interactions, hepatic function, renal function, species, site of infection, therapeutic drug monitoring, and comorbidities.

DPWG

Population: Unspecified **Classification:** No recommendation

Implications: **CYP2C19:*1/*1**
The guideline does not provide a description of the impact of a normal metabolizer phenotype on voriconazole.

Recommendation: The guideline does not provide a recommendation for voriconazole in normal metabolizers.

Citation:

- Clinical Pharmacogenetics Implementation Consortium (CPIC) Guidelines for CYP2C19 and Voriconazole Therapy. Clinical pharmacology and therapeutics. 2017. PMID:27981572
- Pharmacogenetics: from bench to byte--an update of guidelines. Clinical pharmacology and therapeutics. 2011. PMID:21412232

vortioxetine

CPIC

Population: General **Classification:** No recommendation

Implications: **CYP2D6:*49/*36+*10**
CYP2D6: n/a

Recommendation: No recommendation

Citation:

- Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for CYP2D6, CYP2C19, CYP2B6, SLC6A4, and HTR2A Genotypes and Serotonin Reuptake Inhibitor Antidepressants. Clinical pharmacology and therapeutics. 2023. PMID:37032427

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Pharmacogenomics Report

John Doe (4376)

3. Genotype Information

This section provides an overview of the predicted response of the patient. This information is based solely on the genotype information and is not based on a complete patient profile. Detection or absence of variants does not replace the need of therapeutic monitoring. Physicians should consider the information contained in other sections, as well as consider current prescriptions, family history, presenting symptoms, and other factors before making any clinical or therapeutic decisions.

Drugs	Gene	Genotype	Allele Functionality	Phenotypes
rosuvastatin allopurinol	ABCG2	rs2231142 reference (G)/rs2231142 reference (G)	Two Normal function alleles	Normal Function
desflurane enflurane halothane isoflurane methoxyflurane sevoflurane succinylcholine	CACNA1S	Reference/Reference	Two Normal function alleles	Uncertain Susceptibility
ivacaftor	CFTR	Reference/Reference	Two ivacaftor non-responsive alleles	ivacaftor non-responsive in CF patients
efavirenz sertraline	CYP2B6	*1/*1	Two Normal function alleles	Normal Metabolizer
amitriptyline citalopram clomipramine clopidogrel dexlansoprazole doxepin escitalopram imipramine lansoprazole omeprazole pantoprazole sertraline trimipramine voriconazole	CYP2C19	*1/*1	Two Normal function alleles	Normal Metabolizer
celecoxib flurbiprofen fluvastatin fosphenytoin ibuprofen lornoxicam meloxicam phenytoin piroxicam tenoxicam warfarin siponimod	CYP2C9	*1/*1	Two Normal function alleles	Normal Metabolizer

Pharmacogenomics Report

John Doe (4376)

Drugs	Gene	Genotype	Allele Functionality	Phenotypes
amitriptyline atomoxetine clomipramine codeine desipramine doxepin fluvoxamine hydrocodone imipramine nortriptyline ondansetron paroxetine tamoxifen tramadol trimipramine tropisetron venlafaxine vortioxetine	CYP2D6	*49/*36+*10	One Unassigned function allele and one Decreased function allele,Two Unassigned function alleles	n/a
tacrolimus	CYP3A5	*3/*3	Two No function alleles	Poor Metabolizer
warfarin	CYP4F2	*1/*1	N/A	N/A
capecitabine fluorouracil flucytosine tegafur	DPYD	Reference/c.85T>C (*9A)	Two Normal function alleles,One Unassigned function allele and one Normal function allele	Normal Metabolizer,2.0 (Normal Metabolizer)
dapsone methylene blue nitrofurantoin pegloticase primaquine rasburicase tafenoquine toluidine blue	G6PD	B (reference)/B (reference)	Two IV/Normal alleles	Normal
azathioprine mercaptopurine thioguanine	NUDT15	*2/*6	One Uncertain function allele and one No function allele,One Decreased function allele and one No function allele	Possible Intermediate Metabolizer,Poor Metabolizer
desflurane enflurane halothane isoflurane methoxyflurane sevoflurane succinylcholine	RYR1	Reference/Reference	Two Normal function alleles	Uncertain Susceptibility
atorvastatin fluvastatin lovastatin pitavastatin pravastatin rosuvastatin simvastatin	SLCO1B1	*37/*37	Two Normal function alleles,Two Unassigned function alleles	Normal Function,n/a

Drugs	Gene	Genotype	Allele Functionality	Phenotypes
azathioprine mercaptopurine thioguanine	TPMT	*1/*1	Two Normal function alleles	Normal Metabolizer
atazanavir irinotecan	UGT1A1	*1/*6	One Decreased function allele and one Normal function allele	Intermediate Metabolizer
warfarin acenocoumarol phenprocoumon	VKORC1	rs9923231 variant (T)/rs9923231 variant (T)	N/A,Two Higher coumarin sensitivity alleles	N/A,-1639 AA
quetiapine	CYP3A4	*1/*1	Two Normal function alleles	Normal Metabolizer
hormonal contraceptives for systemic use	F5	rs6025 C/rs6025 C	Two Factor V Leiden negative alleles	Factor V Leiden absent

4. Allele Matching Details

ABCG2 allele match data

Genotype matched: rs2231142 reference (G)/rs2231142 reference (G)
Phasing status: Phased

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chr4:88131171	rs2231142	G|G	G	rs2231142 variant (T) - Decreased function	

CACNA1S allele match data

Genotype matched: Reference/Reference
Phasing status: Phased

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chr1:201060815	rs1800559	C|C	C	c.3257G>A - Malignant Hyperthermia associated	
chr1:201091993	rs772226819	G|G	G	c.520C>T - Malignant Hyperthermia associated	

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Pharmacogenomics Report

John Doe (4376)

CFTR allele match data

Genotype matched: Reference/Reference
Phasing status: Phased

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chr7:117509035	rs397508256	G|G	G	E56K - ivacaftor responsive	
chr7:117509069	rs368505753	C|C	C	P67L - ivacaftor responsive	
chr7:117509089	rs115545701	C|C	C	R74W - ivacaftor responsive	
chr7:117530953	rs113993958	G|G	G	D110H - ivacaftor responsive	
chr7:117530955	rs397508537	C|C	C	D110E - ivacaftor responsive	
chr7:117530974	rs77834169	C|C	C	R117C - ivacaftor responsive	
chr7:117530975	rs78655421	G|G	G	R117H - ivacaftor responsive	
chr7:117534318	rs80282562	G|G	G	G178R - ivacaftor responsive	
chr7:117534363	rs397508759	G|G	G	E193K - ivacaftor responsive	
chr7:117534368	rs397508761	A|A	A	711+3A→G - ivacaftor responsive	
chr7:117535285	rs121908752	T|T	T	L206W - ivacaftor responsive	
chr7:117540270	rs77932196	G|G	G	R347H - ivacaftor responsive	
chr7:117540285	rs121908753	G|G	G	R352Q - ivacaftor responsive	
chr7:117548795	rs74551128	C|C	C	A455E - ivacaftor responsive	
chr7:117587799	rs121908757	A|A	A	S549R(A>C) - ivacaftor responsive	
chr7:117587800	rs121908755	G|G	G	S549N - ivacaftor responsive	

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Pharmacogenomics Report

John Doe (4376)

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chr7:117587801	rs121909005	T|T	T	S549R(T>G) - ivacaftor responsive	
chr7:117587805	rs121909013	G|G	G	G551S - ivacaftor responsive	
chr7:117587806	rs75527207	G|G	G	G551D - ivacaftor responsive	
chr7:117590409	rs397508288	A|A	A	D579G - ivacaftor responsive	
chr7:117594930	rs397508387	G|G	G	E831X - ivacaftor responsive	
chr7:117602868	rs80224560	G|G	G	2789+5G→A - ivacaftor responsive	
chr7:117603708	rs397508442	C|C	C	S945L - ivacaftor responsive	
chr7:117606695	rs141033578	C|C	C	S977F - ivacaftor responsive	
chr7:117611555	rs76151804	A|A	A	3272-26A→G - ivacaftor responsive	
chr7:117611595	rs150212784	T|T	T	F1052V - ivacaftor responsive	
chr7:117611620	rs397508513	A|A	A	K1060T - ivacaftor responsive	
chr7:117611640	rs121909020	G|G	G	A1067T - ivacaftor responsive	
chr7:117611646	rs200321110	G|G	G	G1069R - ivacaftor responsive	
chr7:117611649	rs202179988	C|C	C	R1070W - ivacaftor responsive	
chr7:117611650	rs78769542	G|G	G	R1070Q - ivacaftor responsive	
chr7:117611663	rs186045772	T|T	T	F1074L - ivacaftor responsive	
chr7:117614699	rs75541969	G|G	G	D1152H - ivacaftor responsive	
chr7:117639961	rs75039782	C|C	C	3849+10kbC→T - ivacaftor responsive	
chr7:117642451	rs267606723	G|G	G	G1244E - ivacaftor responsive	

Pharmacogenomics Report

John Doe (4376)

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chr7:117642472	rs74503330	G|G	G	S1251N - ivacaftor responsive	
chr7:117642483	rs121909041	T|T	T	S1255P - ivacaftor responsive	
chr7:117642528	rs11971167	G|G	G	D1270N - ivacaftor responsive	
chr7:117664770	rs193922525	G|G	G	G1349D - ivacaftor responsive	

CYP2B6 allele match data

Genotype matched: *1/*1
Phasing status: Phased

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chr19:40991224	rs34223104	T|T	T	*22 - Increased function,*34 - Decreased function,*35 - No function,*36 - Decreased function	
chr19:40991367	rs34883432	A|A	A	*10 - Uncertain function	
chr19:40991369	rs8192709	C|C	C	*2 - Normal function,*10 - Uncertain function	
chr19:40991381	rs33973337	A|A	A	*17 - Normal function	
chr19:40991388	rs33980385	A|A	A	*17 - Normal function	
chr19:40991390	rs33926104	C|C	C	*17 - Normal function	
chr19:40991391	rs34284776	G|G	G	*17 - Normal function	
chr19:40991441	rs35303484	A|A	A	*11 - Uncertain function	
chr19:41004015	rs281864907	T|T	T	*38 - No function	
chr19:41004125	rs36060847	G|G	G	*12 - No function	
chr19:41004133	rs148009906	G|G	G	*44 - Unassigned function	
chr19:41004158	rs186335453	G|G	G	*35 - No function	

Pharmacogenomics Report

John Doe (4376)

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chr19:41004303	rs139801276	T|T	T	*35 - No function	
chr19:41004377	rs12721655	A|A	A	*8 - No function,*13 - No function	
chr19:41004380	rs535039125	C|C	C	*39 - Unassigned function	
chr19:41004381	rs35773040	G|G	G	*14 - Uncertain function	
chr19:41004406	rs145884402	G|G	G	*35 - No function	
chr19:41006919	rs3826711	C|C	C	*26 - Decreased function	
chr19:41006923	rs36056539	C|C	C	*20 - Decreased function	
chr19:41006936	rs3745274	G|G	G	*6 - Decreased function,*7 - Decreased function,*9 - Decreased function,*13 - No function,*19 - Decreased function,*20 - Decreased function,*26 - Decreased function,*34 - Decreased function,*36 - Decreased function,*37 - No function,*38 - No function,*39 - Unassigned function,*40 - Unassigned function,*41 - Unassigned function,*42 - Unassigned function,*43 - Unassigned function	
chr19:41006967	rs58871670	G|G	G	*45 - Unassigned function	
chr19:41006968	rs373489637	T|T	T	*37 - No function	
chr19:41007013	rs36079186	T|T	T	*27 - Uncertain function,*35 - No function	

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Pharmacogenomics Report

John Doe (4376)

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chr19:41009313		A|A	A	*46 - Unassigned function	
chr19:41009350	rs45482602	C|C	C	*3 - Uncertain function	
chr19:41009358	rs2279343	A|A	A	*4 - Increased function,*6 - Decreased function,*7 - Decreased function,*13 - No function,*18 - No function,*19 - Decreased function,*20 - Decreased function,*26 - Decreased function,*34 - Decreased function,*36 - Decreased function,*37 - No function,*38 - No function,*39 - Unassigned function,*40 - Unassigned function,*41 - Unassigned function,*42 - Unassigned function,*43 - Unassigned function	
chr19:41010006	rs139029625	G|G	G	*35 - No function	
chr19:41010088	rs34698757	C|C	C	*28 - No function	
chr19:41010108	rs193922917	C|C	C	*31 - Normal function	
chr19:41012316	rs28399499	T|T	T	*18 - No function	
chr19:41012339	rs34826503	C|C	C	*19 - Decreased function	
chr19:41012393	rs754621576	T|T	T	*47 - Unassigned function	
chr19:41012394	rs780991919	A|A	A	*47 - Unassigned function	
chr19:41012465	rs34097093	C|C	C	*28 - No function	

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Pharmacogenomics Report

John Doe (4376)

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chr19:41012466	rs200458614	G|G	G	*40 - Unassigned function	
chr19:41012471	rs201500445	T|T	T	*41 - Unassigned function	
chr19:41012478	rs200238771	T|T	T	*48 - Unassigned function	
chr19:41012693	rs35979566	T|T	T	*15 - Uncertain function	
chr19:41012740	rs193922918	G|G	G	*32 - Normal function	
chr19:41012803	rs35010098	C|C	C	*21 - Uncertain function	
chr19:41016652	rs764288403	G|G	G	*49 - Unassigned function	
chr19:41016679	rs374099483	G|G	G	*42 - Unassigned function	
chr19:41016726	rs3211369	A|A	A	*23 - Unknown function	
chr19:41016741	rs117872433	G|G	G	*43 - Unassigned function	
chr19:41016778	rs564083989	G|G	G	*24 - No function	
chr19:41016805		A|A	A	*25 - Unknown function	
chr19:41016810	rs3211371	C|C	C	*5 - Normal function,*7 - Decreased function,*33 - Uncertain function,*34 - Decreased function	

CYP2C19 allele match data

Genotype matched: *1/*1
Phasing status: Unphased

Pharmacogenomics Report

John Doe (4376)

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chr10:94761900	rs12248560	C|C	C	*1 - Normal function,*4 - No function,*17 - Increased function	
chr10:94762706	rs28399504	A|A	A	*1 - Normal function,*4 - No function	
chr10:94762712	rs367543002	C|C	C	*1 - Normal function,*34 - Uncertain function	
chr10:94762715	rs367543003	T|T	T	*1 - Normal function,*34 - Uncertain function	
chr10:94762755	rs55752064	T|T	T	*1 - Normal function,*14 - Uncertain function	
chr10:94762760	rs17882687	A|A	A	*1 - Normal function,*15 - Normal function,*28 - Normal function,*35 - No function,*39 - Uncertain function	
chr10:94762788	rs1564656981	A|A	A	*1 - Normal function,*29 - Uncertain function	
chr10:94762856	rs1564657013	A|A	A	*1 - Normal function,*19 - Decreased function	
chr10:94775106	rs145328984	C|C	C	*1 - Normal function,*30 - Uncertain function	
chr10:94775121	rs1564660997	C|C	C	*1 - Normal function,*31 - Uncertain function	
chr10:94775160	rs118203756	G|G	G	*1 - Normal function,*23 - Uncertain function	
chr10:94775185	rs1288601658	A|A	A	*1 - Normal function,*32 - Uncertain function	
chr10:94775367	rs12769205	A|A	A	*1 - Normal function,*2 - No function,*35 - No function	

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Pharmacogenomics Report

John Doe (4376)

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chr10:94775416	rs41291556	T|T	T	*1 - Normal function,*8 - No function	
chr10:94775423	rs17885179	A|A	A	*1 - Normal function,*39 - Uncertain function	
chr10:94775453	rs72552267	G|G	G	*1 - Normal function,*6 - No function	
chr10:94775489	rs17884712	G|G	G	*1 - Normal function,*9 - Decreased function	
chr10:94775507	rs58973490	G|G	G	*1 - Normal function,*2 - No function,*11 - Normal function	
chr10:94780574	rs140278421	G|G	G	*1 - Normal function,*22 - No function	
chr10:94780579	rs370803989	G|G	G	*1 - Normal function,*33 - Uncertain function	
chr10:94780653	rs4986893	G|G	G	*1 - Normal function,*3 - No function	
chr10:94781858	rs6413438	C|C	C	*1 - Normal function,*10 - Decreased function	
chr10:94781859	rs4244285	G|G	G	*1 - Normal function,*2 - No function	
chr10:94781944	rs375781227	G|G	G	*1 - Normal function,*26 - Decreased function	
chr10:94781999	rs72558186	T|T	T	*1 - Normal function,*7 - No function	
chr10:94842861	rs138142612	G|G	G	*1 - Normal function,*18 - Normal function	

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Pharmacogenomics Report

John Doe (4376)

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chr10:94842866	rs3758581	G/G	A	*1 - Normal function,*2 - No function,*3 - No function,*4 - No function,*5 - No function,*6 - No function,*7 - No function,*8 - No function,*9 - Decreased function,*10 - Decreased function,*11 - Normal function,*12 - Uncertain function,*13 - Normal function,*14 - Uncertain function,*15 - Normal function,*17 - Increased function,*18 - Normal function,*19 - Decreased function,*22 - No function,*23 - Uncertain function,*24 - No function,*25 - Decreased function,*26 - Decreased function,*28 - Normal function,*29 - Uncertain function,*31 - Uncertain function,*32 - Uncertain function,*33 - Uncertain function,*35 - No function,*39 - Uncertain function	
chr10:94842879	rs118203757	G|G	G	*1 - Normal function,*24 - No function	
chr10:94842995	rs113934938	G|G	G	*1 - Normal function,*28 - Normal function	

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Pharmacogenomics Report

John Doe (4376)

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chr10:94849995	rs17879685	C|C	C	*1 - Normal function,*13 - Normal function	
chr10:94852738	rs56337013	C|C	C	*1 - Normal function,*5 - No function	
chr10:94852765	rs192154563	C|C	C	*1 - Normal function,*16 - Decreased function	
chr10:94852785	rs118203759	C|C	C	*1 - Normal function,*25 - Decreased function	
chr10:94852914	rs55640102	A|A	A	*1 - Normal function,*12 - Uncertain function	

CYP2C9 allele match data

Genotype matched: *1/*1
Phasing status: Phased

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chr10:94938683	rs114071557	A|A	A	*36 - n/a	
chr10:94938719		T|T	T	*80 - Unassigned function	
chr10:94938737	rs67807361	C|C	C	*7 - n/a	
chr10:94938771	rs142240658	C|C	C	*21 - n/a	
chr10:94938788		C|C	C	*83 - Unassigned function	
chr10:94938800	rs1364419386	G|G	G	*76 - Unassigned function	
chr10:94938803	rs2031308986	A|A	A	*22 - n/a	
chr10:94938828	rs564813580	A|A	A	*37 - 0.5	
chr10:94941897	rs371055887	G|G	G	*20 - n/a	
chr10:94941915		G|G	G	*23 - 0.5	
chr10:94941958	rs72558187	T|T	T	*13 - 0.0	

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Pharmacogenomics Report

John Doe (4376)

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chr10:94941975		G|G	G	*77 - Unassigned function	
chr10:94941976		G|G	G	*38 - 0.5	
chr10:94941982	rs762239445	G|G	G	*39 - 0.0	
chr10:94942018		T|T	T	*40 - n/a	
chr10:94942205	rs1304490498	CAATGGAAAGA|CAATGGAAAGA	CAATGGAAAGA	*25 - 0.0	
chr10:94942216	rs774607211	A|A	A	*41 - n/a	
chr10:94942230	rs767576260	C|C	C	*43 - 0.0	
chr10:94942231	rs12414460	G|G	G	*42 - 0.0	
chr10:94942233	rs375805362	C|C	C	*62 - n/a	
chr10:94942234	rs72558189	G|G	G	*14 - 0.5,*35 - 0.0	
chr10:94942243	rs1375956433	T|T	T	*78 - Unassigned function	
chr10:94942249	rs200965026	C|C	C	*26 - 0.5,*44 - 0.5	
chr10:94942254	rs199523631	C|C	C	*45 - 0.0	
chr10:94942255	rs200183364	G|G	G	*33 - 0.0	
chr10:94942290	rs1799853	C|C	C	*2 - 0.5,*35 - 0.0,*61 - 0.5	
chr10:94942291	rs141489852	G|G	G	*63 - n/a	
chr10:94942305	rs754487195	G|G	G	*46 - 0.5	
chr10:94942306	rs1289704600	C|C	C	*72 - n/a	
chr10:94942308	rs17847037	C|C	C	*73 - n/a	
chr10:94942309	rs7900194	G|G	G	*8 - 0.5,*27 - n/a	
chr10:94947782	rs72558190	C|C	C	*15 - 0.0	
chr10:94947785	rs774550549	C|C	C	*47 - n/a	
chr10:94947869		A|A	A	*69 - n/a	
chr10:94947907		A|A	A	*57 - n/a	
chr10:94947917	rs1326630788	T|T	T	*48 - n/a	
chr10:94947938	rs2031531005	A|A	A	*28 - 0.5	
chr10:94947939	rs370100007	G|G	G	*74 - n/a	

Pharmacogenomics Report

John Doe (4376)

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chr10:94949129		A|A	A	*49 - n/a	
chr10:94949144		C|C	C	*50 - 0.5	
chr10:94949145	rs772782449	C|C	C	*82 - Unassigned function	
chr10:94949161		AT|AT	AT	*85 - Unassigned function	
chr10:94949217	rs2256871	A|A	A	*9 - 1.0	
chr10:94949280	rs9332130	A|A	A	*10 - n/a,*71 - n/a	
chr10:94949281	rs9332131	GA|GA	GA	*6 - 0.0	
chr10:94972119	rs182132442	C|C	C	*29 - 0.5	
chr10:94972123		C|C	C	*64 - n/a	
chr10:94972134		A|A	A	*51 - n/a	
chr10:94972179	rs72558192	A|A	A	*16 - 0.5	
chr10:94972180	rs988617574	C|C	C	*52 - 0.0	
chr10:94972183		A|A	A	*81 - Unassigned function	
chr10:94972233	rs1237225311	C|C	C	*53 - n/a	
chr10:94981199		G|G	G	*65 - n/a	
chr10:94981201	rs57505750	T|T	T	*31 - 0.5	
chr10:94981224	rs28371685	C|C	C	*11 - 0.5	
chr10:94981225	rs367826293	G|G	G	*34 - n/a	
chr10:94981230	rs1274535931	C|C	C	*58 - n/a	
chr10:94981250	rs750820937	C|C	C	*54 - n/a	
chr10:94981258	rs1297714792	C|C	C	*79 - Unassigned function	
chr10:94981281	rs749060448	G|G	G	*24 - 0.0	
chr10:94981296	rs1057910	A|A	A	*3 - 0.0,*18 - n/a,*68 - n/a	
chr10:94981297	rs56165452	T|T	T	*4 - 0.5	
chr10:94981301	rs28371686	C|C	C	*5 - 0.5	
chr10:94981302	rs1250577724	C|C	C	*55 - 0.5	
chr10:94981305	rs578144976	C|C	C	*66 - n/a	

Pharmacogenomics Report

John Doe (4376)

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chr10:94981365		C|C	C	*17 - n/a	
chr10:94981371	rs542577750	G|G	G	*68 - n/a	
chr10:94986042	rs764211126	A|A	A	*56 - n/a	
chr10:94986073	rs72558193	A|A	A	*18 - n/a	
chr10:94986136	rs1254213342	A|A	A	*75 - n/a	
chr10:94986174	rs1441296358	G|G	G	*84 - Unassigned function	
chr10:94988852	rs776908257	C|C	C	*67 - n/a	
chr10:94988855		A|A	A	*59 - n/a	
chr10:94988880		G|G	G	*70 - n/a	
chr10:94988917	rs769942899	G|G	G	*19 - n/a	
chr10:94988925	rs202201137	A|A	A	*61 - 0.5	
chr10:94988955	rs767284820	T|T	T	*60 - n/a	
chr10:94988984	rs781583846	G|G	G	*30 - 0.5	
chr10:94989020	rs9332239	C|C	C	*12 - 0.5,*71 - n/a	
chr10:94989023	rs868182778	G|G	G	*32 - n/a	

CYP2D6 allele match data

Genotype matched: *49/*36+*10
Phasing status: Unphased

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings

CYP3A4 allele match data

Genotype matched: *1/*1
Phasing status: Phased

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chr7:99758183	rs67666821	G|G	G	*20 - Unassigned function	

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Pharmacogenomics Report

John Doe (4376)

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chr7:99758228		T|T	T	*46 - Unassigned function	
chr7:99760836	rs4986913	G|G	G	*19 - Unassigned function	
chr7:99760901	rs4986910	A|A	A	*3 - Unassigned function,*37 - Unassigned function,*38 - Unassigned function	
chr7:99760956	rs774109750	T|T	T	*34 - Unassigned function	
chr7:99762047	rs4986909	G|G	G	*13 - Unassigned function	
chr7:99762054		A|A	A	*45 - Unassigned function	
chr7:99762069		T|T	T	*47 - Unassigned function	
chr7:99762177	rs12721629	G|G	G	*12 - Unassigned function	
chr7:99762186	rs756833413	C|C	C	*33 - Unassigned function	
chr7:99762206	rs67784355	G|G	G	*11 - Unassigned function,*38 - Unassigned function	
chr7:99762234		C|C	C	*48 - Unassigned function	
chr7:99763877	rs368296206	A|A	A	*32 - Unassigned function	
chr7:99763909	rs1303250043	G|G	G	*31 - Unassigned function	
chr7:99763925		T|T	T	*21 - Unassigned function	
chr7:99764003	rs28371759	A|A	A	*18 - Unassigned function	
chr7:99766411	rs4646438	G|G	G	*6 - Unassigned function	
chr7:99766424		T|T	T	*44 - Unassigned function	
chr7:99766439		C|C	C	*43 - Unassigned function	

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Pharmacogenomics Report

John Doe (4376)

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chr7:99766440	rs138105638	G|G	G	*26 - Unassigned function	
chr7:99768360	rs55785340	A|A	A	*2 - Unassigned function	
chr7:99768371	rs55901263	G|G	G	*5 - Unassigned function	
chr7:99768424	rs113667357	T|T	T	*24 - Unassigned function	
chr7:99768447		T|T	T	*42 - Unassigned function	
chr7:99768458	rs4987161	A|A	A	*17 - Unassigned function	
chr7:99768470	rs12721627	G|G	G	*16 - Unassigned function	
chr7:99768693	rs35599367	G|G	G	*22 - Unassigned function,*37 - Unassigned function	
chr7:99769769	rs4986908	C|C	C	*10 - Unassigned function	
chr7:99769781	rs72552798	C|C	C	*9 - Unassigned function	
chr7:99769804	rs4986907	C|C	C	*15 - Unassigned function	
chr7:99769805	rs57409622	G|G	G	*23 - Unassigned function	
chr7:99770165	rs72552799	C|C	C	*8 - Unassigned function	
chr7:99770166	rs778013004	G|G	G	*30 - Unassigned function	
chr7:99770196		T|T	T	*41 - Unassigned function	
chr7:99770202	rs55951658	T|T	T	*4 - Unassigned function	
chr7:99770217	rs1449865051	A|A	A	*29 - Unassigned function	
chr7:99778079	rs56324128	C|C	C	*7 - Unassigned function	
chr7:99780036		G|G	G	*40 - Unassigned function	

Pharmacogenomics Report

John Doe (4376)

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chr7:99784018	rs570051168	G|G	G	*28 - Unassigned function	
chr7:99784038	rs12721634	A|A	A	*14 - Unassigned function	
chr7:99784075	rs188389063	G|G	G	*35 - Unassigned function	
chr7:99784078		C|C	C	*39 - Unassigned function	

CYP3A5 allele match data

Genotype matched: *3/*3
Phasing status: Unphased

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chr7:99652770	rs41303343	T|T	T	*7 - No function	
chr7:99660516	rs28383479	C|C	C	*9 - Unknown function	
chr7:99665212	rs10264272	C|C	C	*6 - No function	
chr7:99672916	rs776746	C/C	T	*3 - No function	
chr7:99676198	rs55817950	G|G	G	*8 - Unknown function	

CYP4F2 allele match data

Genotype matched: *1/*1
Phasing status: Phased

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chr19:15878779	rs3093200	G|G	G	*5 - Unassigned function	
chr19:15879412	rs138971789	C|C	C	*15 - Unassigned function	
chr19:15879621	rs2108622	C|C	C	*3 - Unassigned function,*4 - Unassigned function	

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Pharmacogenomics Report

John Doe (4376)

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chr19:15886018	rs145174239	G|G	G	*14 - Unassigned function	
chr19:15889671	rs144233412	C|C	C	*13 - Unassigned function	
chr19:15890405	rs3093153	C|C	C	*6 - Unassigned function	
chr19:15892541	rs145875499	C|C	C	*12 - Unassigned function	
chr19:15895527	rs114396708	G|G	G	*11 - Unassigned function	
chr19:15895560	rs144455532	G|G	G	*10 - Unassigned function	
chr19:15897466	rs201380574	C|C	C	*9 - Unassigned function	
chr19:15897473	rs115517770	G|G	G	*8 - Unassigned function	
chr19:15897566	rs114099324	C|C	C	*7 - Unassigned function	
chr19:15897578	rs3093105	A|A	A	*2 - Unassigned function,*4 - Unassigned function	

DPYD allele match data

Genotype matched: Reference/c.85T>C (*9A)
Phasing status: Unphased

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chr1:97078987	rs114096998	G|G	G	c.3067C>A - 1.0	
chr1:97078993	rs148799944	C|C	C	c.3061G>C - 1.0	
chr1:97079005	rs140114515	C|C	C	c.3049G>A - 1.0	
chr1:97079071	rs1801268	C|C	C	c.2983G>T (*10) - 0.0	
chr1:97079076	rs139459586	A|A	A	c.2978T>G - 1.0	
chr1:97079077	rs202144771	G|G	G	c.2977C>T - 1.0	
chr1:97079121	rs72547601	T|T	T	c.2933A>G - 0.0	

Pharmacogenomics Report

John Doe (4376)

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chr1:97079133	rs72547602	T|T	T	c.2921A>T - 1.0	
chr1:97079139	rs145529148	T|T	T	c.2915A>G - 1.0	
chr1:97082365	rs141044036	T|T	T	c.2872A>G - 0.0	
chr1:97082391	rs67376798	T|T	T	c.2846A>T - 0.5	
chr1:97098598	rs1801267	C|C	C	c.2657G>A (*9B) - 1.0	
chr1:97098599	rs147545709	G|G	G	c.2656C>T - 1.0	
chr1:97098616	rs55674432	C|C	C	c.2639G>T - 0.0	
chr1:97098632	rs201035051	T|T	T	c.2623A>C - 1.0	
chr1:97193109	rs60139309	T|T	T	c.2582A>G - 1.0	
chr1:97193209	rs200687447	C|C	C	c.2482G>A - 1.0	
chr1:97234958	rs199634007	G|G	G	c.2336C>A - 1.0	
chr1:97234991	rs56005131	G|G	G	c.2303C>A - 1.0	
chr1:97305279	rs112766203	G|G	G	c.2279C>T - 0.5	
chr1:97305363	rs60511679	A|A	A	c.2195T>G - 1.0	
chr1:97305364	rs1801160	C|C	C	c.2194G>A (*6) - 1.0	
chr1:97305372	rs146529561	G|G	G	c.2186C>T - 1.0	
chr1:97306195	rs145548112	C|C	C	c.2161G>A - 1.0	
chr1:97373598	rs137999090	C|C	C	c.2021G>A - 0.0	
chr1:97373629	rs138545885	C|C	C	c.1990G>T - 1.0	
chr1:97382461	rs55971861	T|T	T	c.1906A>C - 1.0	
chr1:97450058	rs3918290	C|C	C	c.1905+1G>A (*2A) - 0.0	
chr1:97450059	rs3918289	G|G	G	c.1905C>G - 1.0	
chr1:97450065	rs72549303	TG|TG	TG	c.1898delC (*3) - 0.0	
chr1:97450068	rs17376848	A|A	A	c.1896T>C - 1.0	
chr1:97450168	rs147601618	A|A	A	c.1796T>C - 1.0	
chr1:97450187	rs145773863	C|C	C	c.1777G>A - 0.0	
chr1:97450189	rs138616379	C|C	C	c.1775G>A - 0.0	
chr1:97450190	rs59086055	G|G	G	c.1774C>T - 0.0	

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Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chr1:97515784	rs201615754	C|C	C	c.1682G>T - 1.0	
chr1:97515787	rs55886062	A|A	A	c.1679T>G (*13) - 0.0	
chr1:97515839	rs1801159	T|T	T	c.1627A>G (*5) - 1.0	
chr1:97515851	rs142619737	C|C	C	c.1615G>A - 1.0	
chr1:97515865	rs1801158	C|C	C	c.1601G>A (*4) - 1.0	
chr1:97515889	rs190951787	G|G	G	c.1577C>G - 1.0	
chr1:97515923	rs148994843	C|C	C	c.1543G>A - 1.0	
chr1:97549565	rs138391898	C|C	C	c.1519G>A - 1.0	
chr1:97549600	rs111858276	T|T	T	c.1484A>G - 0.0	
chr1:97549609	rs72549304	G|G	G	c.1475C>T - 0.0	
chr1:97549681	rs199549923	G|G	G	c.1403C>A - 1.0	
chr1:97549713	rs57918000	G|G	G	c.1371C>T - 1.0	
chr1:97549726	rs144395748	G|G	G	c.1358C>G - 1.0	
chr1:97549735	rs72975710	G|G	G	c.1349C>T - 1.0	
chr1:97573785	rs186169810	A|A	A	c.1314T>G - 0.5	
chr1:97573805	rs142512579	C|C	C	c.1294G>A - 1.0	
chr1:97573821	rs764666241	C|C	C	c.1278G>T - 1.0	
chr1:97573839	rs200064537	A|A	A	c.1260T>A - 1.0	
chr1:97573863	rs56038477	C|C	C	c.1129-5923C>G, c.1236G>A (HapB3) - 0.5	
chr1:97573881	rs61622928	C|C	C	c.1218G>A - 1.0	
chr1:97573918	rs143815742	C|C	C	c.1181G>T - 1.0	
chr1:97573919	rs140602333	G|G	G	c.1180C>T - 1.0	
chr1:97573943	rs78060119	C|C	C	c.1156G>T (*12) - 0.0	
chr1:97579893	rs75017182	G|G	G	c.1129-5923C>G, c.1236G>A (HapB3) - 0.5	
chr1:97593238	rs72549305	T|T	T	c.1108A>G - 1.0	
chr1:97593289	rs143154602	G|G	G	c.1057C>T - 0.0	
chr1:97593322	rs183385770	C|C	C	c.1024G>A - 0.0	

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Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chr1:97593343	rs72549306	C|C	C	c.1003G>T (*11) - 1.0	
chr1:97593379	rs201018345	C|C	C	c.967G>A - 1.0	
chr1:97595083	rs145112791	G|G	G	c.934C>T - 1.0	
chr1:97595088	rs150437414	A|A	A	c.929T>C - 1.0	
chr1:97595149	rs146356975	T|T	T	c.868A>G - 0.5	
chr1:97679170	rs45589337	T|T	T	c.775A>G - 1.0	
chr1:97691776	rs1801266	G|G	G	c.703C>T (*8) - 0.0	
chr1:97699399	rs72549307	T|T	T	c.632A>G - 0.0	
chr1:97699430	rs72549308	T|T	T	c.601A>C - 0.0	
chr1:97699474	rs115232898	T|T	T	c.557A>G - 0.5	
chr1:97699506	rs6670886	C|C	C	c.525G>A - 1.0	
chr1:97699533	rs139834141	C|C	C	c.498G>A - 1.0	
chr1:97699535	rs2297595	T|T	T	c.496A>G - 1.0	
chr1:97721542	rs200562975	T|T	T	c.451A>G - 1.0	
chr1:97721650	rs141462178	T|T	T	c.343A>G - 1.0	
chr1:97740400	rs150385342	C|C	C	c.313G>A - 1.0	
chr1:97740410	rs72549309	GATGA|GATGA	GATGA	c.295_298delTCAT (*7) - 0.0	
chr1:97883329	rs1801265	A/G	A	c.85T>C (*9A) - 1.0	
chr1:97883352	rs80081766	C|C	C	c.62G>A - 1.0	
chr1:97883353	rs72549310	G|G	G	c.61C>T - 0.0	
chr1:97883368	rs150036960	G|G	G	c.46C>G - 1.0	

F5 allele match data

Genotype matched: rs6025 C/rs6025 C
Phasing status: Phased

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chr1:169549811	rs6025	C|C	C	rs6025 T (Factor V Leiden) - Unassigned function	

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G6PD allele match data

Genotype matched: B (reference)/B (reference)
Phasing status: Phased

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chrX:154532046		A|A	A	Bangkok Noi - I/Deficient with CNSHA	
chrX:154532055		CTCT|CTCT	CTCT	Brighton - I/Deficient with CNSHA	
chrX:154532082		G|G	G	Arakawa - I/Deficient with CNSHA	
chrX:154532083		G|G	G	Buenos Aires - I/Deficient with CNSHA	
chrX:154532085		C|C	C	Campinas - I/Deficient with CNSHA	
chrX:154532086		C|C	C	Fukaya - I/Deficient with CNSHA	
chrX:154532203	rs137852348	G|G	G	Split - III/Deficient	
chrX:154532231		T|T	T	Laibin - Uncertain function	
chrX:154532245	rs137852344	G|G	G	Neapolis - III/Deficient	
chrX:154532257	rs72554664	C|C	C	Kaiping, Anant, Dhon, Sapporo-like, Wosera - II/Deficient	
chrX:154532258		G|G	G	Flores - II/Deficient,Kamiube, Keelung - III/Deficient	
chrX:154532264	rs782608284	C|C	C	Yunan - Uncertain function	
chrX:154532265		C|C	C	Nice - III/Deficient	

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Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chrX:154532269	rs72554665	C|C	C	Bangkok Noi - I/Deficient with CNSHA,Canton, Taiwan-Hakka, Gifu-like, Agrigento-like - II/Deficient,Cosenza - II/Deficient	
chrX:154532278		T|T	T	Amiens - I/Deficient with CNSHA	
chrX:154532279		C|C	C	Figuera da Foz - I/Deficient with CNSHA	
chrX:154532389	rs137852324	C|C	C	Andalus - II/Deficient	
chrX:154532390	rs398123546	G|G	G	Hermoupolis - II/Deficient,Honiara - I/Deficient with CNSHA,Union,Maewo, Chinese-2, Kalo - II/Deficient	
chrX:154532392		A|A	A	Harima - I/Deficient with CNSHA	
chrX:154532403		C|C	C	Cassano - II/Deficient,Hermoupolis - II/Deficient	
chrX:154532408		T|T	T	S. Antioco - II/Deficient	
chrX:154532411	rs137852317	C|C	C	Santiago de Cuba, Morioka - I/Deficient with CNSHA	
chrX:154532432		G|G	G	Telti, Kobe - I/Deficient with CNSHA	
chrX:154532434	rs137852337	C|C	C	Pawnee - II/Deficient	
chrX:154532458		A|A	A	Sumare - I/Deficient with CNSHA	
chrX:154532459	rs782098548	C|C	C	Surabaya - II/Deficient	
chrX:154532570		G|G	G	Georgia - I/Deficient with CNSHA	
chrX:154532590		G|G	G	202G>A_376A>G_1264C>G - I/Deficient with CNSHA	

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John Doe (4376)

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chrX:154532608		C|C	C	Tokyo, Fukushima - I/Deficient with CNSHA	
chrX:154532623		T|T	T	Munich - I/Deficient with CNSHA	
chrX:154532625	rs137852336	C|C	C	Japan, Shinagawa - I/Deficient with CNSHA,Kawasaki - I/Deficient with CNSHA	
chrX:154532626	rs137852323	C|C	C	Riverside - I/Deficient with CNSHA	
chrX:154532628		G|G	G	Suwalki - I/Deficient with CNSHA	
chrX:154532629		G|G	G	Utrecht - I/Deficient with CNSHA	
chrX:154532634		T|T	T	Abeno - II/Deficient	
chrX:154532639		C|C	C	Clinic - I/Deficient with CNSHA	
chrX:154532649		G|G	G	Covao do Lobo - I/Deficient with CNSHA	
chrX:154532661		T|T	T	Anadia - II/Deficient	
chrX:154532662	rs137852325	C|C	C	Puerto Limon - I/Deficient with CNSHA	
chrX:154532667		G|G	G	Bari - I/Deficient with CNSHA	
chrX:154532674	rs137852335	C|C	C	Alhambra - I/Deficient with CNSHA	
chrX:154532676	rs137852316	C|C	C	Nashville, Anaheim, Portici - I/Deficient with CNSHA	
chrX:154532677		G|G	G	Wisconsin - I/Deficient with CNSHA	
chrX:154532679		A|A	A	Krakow - I/Deficient with CNSHA	
chrX:154532688		T|T	T	Praha - I/Deficient with CNSHA	

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Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chrX:154532692		T|T	T	Hartford - I/Deficient with CNSHA	
chrX:154532694	rs137852321	C|C	C	Beverly Hills, Genova, Iwate, Niigata, Yamaguchi - I/Deficient with CNSHA	
chrX:154532695	rs137852334	G|G	G	Guadalajara - I/Deficient with CNSHA,Mt Sinai - I/Deficient with CNSHA	
chrX:154532698	rs137852320	T|T	T	Iowa, Walter Reed, Springfield - I/Deficient with CNSHA	
chrX:154532699		G|G	G	Madrid - I/Deficient with CNSHA	
chrX:154532700		C|C	C	Lynwood - I/Deficient with CNSHA	
chrX:154532701	rs137852322	A|A	A	Tomah - I/Deficient with CNSHA	
chrX:154532713		A|A	A	Olomouc - I/Deficient with CNSHA	
chrX:154532715		A|A	A	Riley - I/Deficient with CNSHA	
chrX:154532716		T|T	T	Calvo Mackenna - I/Deficient with CNSHA	
chrX:154532722	rs371489738	C|C	C	Montpellier - I/Deficient with CNSHA	
chrX:154532752		CGGCCTTGCGCTCGTTCAG|CGGCCTTGCGCTCGTTCAG	CGGCCTTGCGCTCGTTCAG	Tondela - I/Deficient with CNSHA	
chrX:154532758		T|T	T	Tenri - I/Deficient with CNSHA	
chrX:154532765	rs137852329	G|G	G	Aachen - I/Deficient with CNSHA,Loma Linda - I/Deficient with CNSHA	
chrX:154532772	rs137852345	G|G	G	Serres - I/Deficient with CNSHA	

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Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chrX:154532773		C|C	C	Iwatsuki - I/Deficient with CNSHA	
chrX:154532797	rs137852333	G|G	G	Ierapetra - II/Deficient	
chrX:154532802		C|C	C	Partenope - II/Deficient	
chrX:154532945	rs34193178	C|C	C	Mira d'Aire - IV/Normal	
chrX:154532956	rs398123544	T|T	T	Cincinnati - I/Deficient with CNSHA	
chrX:154532969	rs137852342	G|G	G	Chinese-5 - III/Deficient	
chrX:154532987		T|T	T	Torun - I/Deficient with CNSHA	
chrX:154532989		G|G	G	Fushan - II/Deficient	
chrX:154532990	rs5030869	C|C	C	Chatham - II/Deficient	
chrX:154533004		C|C	C	Insuli - IV/Normal	
chrX:154533012		CGTGGGGTCGTCCAGGTACCCTTTG|CGTGGGGTCGTCCAGGTACCCTTTG	CGTGGGGTCGTCCAGGTACCCTTTG	Nara - I/Deficient with CNSHA	
chrX:154533016		G|G	G	Farroupilha - II/Deficient	
chrX:154533025	rs76723693	A|A	A	A- 968C_376G - III/Deficient	
chrX:154533029	rs137852347	A|A	A	Rehevot - I/Deficient with CNSHA	
chrX:154533031		C|C	C	Manhattan - I/Deficient with CNSHA	
chrX:154533044	rs137852339	C|C	C	Kalyan-Kerala, Jamnaga, Rohini - III/Deficient	
chrX:154533064		C|C	C	Ludhiana - II/Deficient	
chrX:154533072		C|C	C	Omiya - I/Deficient with CNSHA	

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Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chrX:154533077		C|C	C	Seoul - II/Deficient	
chrX:154533083		C|C	C	West Virginia - I/Deficient with CNSHA	
chrX:154533122	rs137852327	C|C	C	Ananindeua - II/Deficient,Hechi - II/Deficient,Viangchan, Jammu - II/Deficient	
chrX:154533586	rs74575103	C|C	C	Montalbano - III/Deficient	
chrX:154533587		G|G	G	Osaka - II/Deficient	
chrX:154533589		A|A	A	Piotrkow - I/Deficient with CNSHA	
chrX:154533591		G|G	G	Papua - Uncertain function	
chrX:154533592		T|T	T	Mizushima - II/Deficient	
chrX:154533596	rs137852318	C|C	C	Bajo Maumere - III/Deficient,Seattle, Lodi, Modena, Ferrara II, Athens-like - III/Deficient	
chrX:154533605		T|T	T	Chinese-1 - II/Deficient,Haikou - II/Deficient	
chrX:154533607		G|G	G	Wexham - I/Deficient with CNSHA	
chrX:154533608		A|A	A	La Jolla - I/Deficient with CNSHA	
chrX:154533614		G|G	G	Sugao - I/Deficient with CNSHA	
chrX:154533615		C|C	C	Bangkok - I/Deficient with CNSHA	
chrX:154533619		T|T	T	Lille - I/Deficient with CNSHA	
chrX:154533620		C|C	C	Cleveland Corum - I/Deficient with CNSHA	
chrX:154533629		C|C	C	Roubaix - II/Deficient	

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Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chrX:154533634	rs137852346	C|C	C	Aveiro - I/Deficient with CNSHA	
chrX:154534036		G|G	G	Wayne - I/Deficient with CNSHA	
chrX:154534074		TCAGTGC|TCAGTGC	TCAGTGC	Stonybrook - I/Deficient with CNSHA	
chrX:154534092		T|T	T	Durham - I/Deficient with CNSHA	
chrX:154534102	rs782757170	G|G	G	Nanning - III/Deficient	
chrX:154534110		C|C	C	Asahikawa - I/Deficient with CNSHA	
chrX:154534116		ATGT|ATGT	ATGT	North Dallas - I/Deficient with CNSHA	
chrX:154534125	rs137852328	C|C	C	A- 680T_376G - III/Deficient,Mexico City - III/Deficient	
chrX:154534126		G|G	G	Radlowo - II/Deficient	
chrX:154534157	rs137852319	A|A	A	Harilaou - I/Deficient with CNSHA	
chrX:154534345	rs137852326	C|C	C	Cincinnati - I/Deficient with CNSHA,Minnesota, Marion, Gastonia, LeJeune - I/Deficient with CNSHA	
chrX:154534348	rs782754619	T|T	T	Sibari - III/Deficient	
chrX:154534387	rs781865768	T|T	T	Dagua - Uncertain function	
chrX:154534389	rs137852332	C|C	C	Nilgiri - II/Deficient,Santiago - I/Deficient with CNSHA	
chrX:154534390	rs137852330	G|G	G	Coimbra Shunde - II/Deficient,Vancouver - I/Deficient with CNSHA	

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Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chrX:154534409		G|G	G	Pedoplis-Ckaro - I/Deficient with CNSHA	
chrX:154534414		GGGA|GGGA	GGGA	Tsukui - I/Deficient with CNSHA	
chrX:154534419	rs5030868	G|G	G	Mediterranean, Dallas, Panama, Sassari, Cagliari, Birmingham - II/Deficient	
chrX:154534438	rs267606836	G|G	G	Vancouver - I/Deficient with CNSHA	
chrX:154534440	rs5030872	T|T	T	Malaga - III/Deficient,Santa Maria - II/Deficient	
chrX:154534447		T|T	T	Chikugo - I/Deficient with CNSHA	
chrX:154534455		T|T	T	Shinshu - I/Deficient with CNSHA	
chrX:154534463		G|G	G	Miaoli - II/Deficient	
chrX:154534465	rs137852343	A|A	A	Nankang - II/Deficient	
chrX:154534468		G|G	G	Volendam - I/Deficient with CNSHA	
chrX:154534485		C|C	C	Naone - II/Deficient	
chrX:154534486		G|G	G	Toledo - II/Deficient	
chrX:154534489	rs137852331	T|T	T	Taipei, Chinese-3 - II/Deficient	
chrX:154534494		C|C	C	Plymouth - I/Deficient with CNSHA	
chrX:154534495	rs137852314	C|C	C	Mahidol - III/Deficient	
chrX:154535176	rs370918918	C|C	C	Gond - Uncertain function	
chrX:154535180	rs782487723	C|C	C	Shenzen - II/Deficient	
chrX:154535187	rs137852313	C|C	C	Ilesha - III/Deficient	

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John Doe (4376)

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chrX:154535190		G|G	G	Acrokorinthos - II/Deficient	
chrX:154535211		C|C	C	Liuzhou - II/Deficient	
chrX:154535244		G|G	G	Belem - II/Deficient	
chrX:154535247		G|G	G	Valladolid - II/Deficient	
chrX:154535249	rs782322505	T|T	T	Cairo - II/Deficient	
chrX:154535261		C|C	C	Quing Yan - III/Deficient	
chrX:154535269		G|G	G	Crispim - II/Deficient	
chrX:154535270	rs78365220	A|A	A	Crispim - II/Deficient,Salerno Pyrgos - III/Deficient,Vanua Lava - II/Deficient	
chrX:154535274		C|C	C	Crispim - II/Deficient	
chrX:154535277	rs1050829	T|T	T	202G>A_376A>G_1264C>G - I/Deficient with CNSHA,A - IV/Normal,A-202A_376G - III/Deficient,A-680T_376G - III/Deficient,A-968C_376G - III/Deficient,Acrokorinthos - II/Deficient,Ananindeua - II/Deficient,Mt Sinai - I/Deficient with CNSHA,Santa Maria - II/Deficient,Sierra Leone - III/Deficient	
chrX:154535278		C|C	C	Crispim - II/Deficient	
chrX:154535301		A|A	A	Bao Loc - II/Deficient	
chrX:154535316	rs5030870	C|C	C	Sao Borja - IV/Normal	
chrX:154535330		A|A	A	Hammersmith - III/Deficient	
chrX:154535336	rs267606835	G|G	G	Vancouver - I/Deficient with CNSHA	

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Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chrX:154535342	rs181277621	C|C	C	Sierra Leone - III/Deficient	
chrX:154535367		GCTT|GCTT	GCTT	Urayasu - I/Deficient with CNSHA	
chrX:154535379		G|G	G	Guangzhou - III/Deficient	
chrX:154535962	rs782308266	C|C	C	Lagosanto - III/Deficient	
chrX:154535963	rs138687036	G|G	G	Ube Konan - III/Deficient	
chrX:154535980		A|A	A	Swansea - I/Deficient with CNSHA	
chrX:154535995	rs782090947	T|T	T	Murcia Oristano - III/Deficient	
chrX:154535996	rs137852349	A|A	A	Namouru - II/Deficient	
chrX:154536002	rs1050828	C|C	C	202G>A_376A>G_1264C>G - I/Deficient with CNSHA,A-202A_376G - III/Deficient,Asahi - III/Deficient,Hechi - II/Deficient	
chrX:154536008		A|A	A	Songklanagarind - II/Deficient	
chrX:154536019		G|G	G	Amazonia - II/Deficient,Musashino - III/Deficient	
chrX:154536021		CAGA|CAGA	CAGA	Amsterdam - I/Deficient with CNSHA	
chrX:154536025		A|A	A	Costanzo - II/Deficient	
chrX:154536032	rs137852315	C|C	C	Metaponto - III/Deficient	
chrX:154536034		C|C	C	Palestrina - III/Deficient	
chrX:154536035		G|G	G	Kamogawa - II/Deficient	

Pharmacogenomics Report

John Doe (4376)

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chrX:154536045		C|C	C	Kozukata - I/Deficient with CNSHA	
chrX:154536151		G|G	G	Kambos - III/Deficient	
chrX:154536156	rs76645461	A|A	A	Aures - III/Deficient	
chrX:154536168	rs78478128	G|G	G	Orissa - III/Deficient	
chrX:154536169		C|C	C	Rignano - III/Deficient	
chrX:154546045	rs137852338	CATG|CATG	CATG	Sunderland - I/Deficient with CNSHA	
chrX:154546046		A|A	A	Gidra - Uncertain function	
chrX:154546057		T|T	T	Honiara - I/Deficient with CNSHA	
chrX:154546061	rs137852340	T|T	T	Gaohe - III/Deficient	
chrX:154546116		C|C	C	Lages - III/Deficient	
chrX:154546122		C|C	C	Sinnai - III/Deficient	
chrX:154546131		G|G	G	No name - I/Deficient with CNSHA	

NUDT15 allele match data

Genotype matched: *2/*6
Phasing status: Unphased

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chr13:48037748	rs769369441	T|T	T	*10 - Unassigned function	
chr13:48037749		G|G	G	*19 - Unassigned function	
chr13:48037782	rs746071566	AGGAGTCGGAGTC/AGGAGTCGGAGTC	AGGAGTC	*2 - No function,*6 - Uncertain function,*9 - No function	
chr13:48037798	rs186364861	G|G	G	*5 - Uncertain function	

Pharmacogenomics Report

John Doe (4376)

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chr13:48037825	rs777311140	C|C	C	*14 - Unassigned function	
chr13:48037834	rs1202487323	C|C	C	*16 - Unassigned function	
chr13:48037847	rs766023281	G|G	G	*7 - Uncertain function	
chr13:48037849		A|A	A	*8 - Uncertain function	
chr13:48037885	rs1950545307	G|G	G	*11 - Unassigned function	
chr13:48037902	rs149436418	C|C	C	*12 - Unassigned function	
chr13:48040977	rs1457579126	GA|GA	GA	*18 - Unassigned function	
chr13:48041103	rs761191455	T|T	T	*13 - Unassigned function	
chr13:48041113	rs1368252918	G|G	G	*17 - Unassigned function	
chr13:48045690	rs768324690	C|C	C	*20 - Unassigned function	
chr13:48045719	rs116855232	C/T	C	*2 - No function,*3 - No function	
chr13:48045720	rs147390019	G|G	G	*4 - Uncertain function	
chr13:48045771	rs139551410	T|T	T	*15 - Unassigned function	

RYR1 allele match data

Genotype matched: Reference/Reference
Phasing status: Phased

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chr19:38440802	rs193922747	T|T	T	c.103T>C - Malignant Hyperthermia associated	

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Pharmacogenomics Report

John Doe (4376)

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chr19:38440829	rs193922748	C|C	C	c.130C>T - Malignant Hyperthermia associated	
chr19:38444211	rs118192161	C|C	C	c.487C>T - Malignant Hyperthermia associated	
chr19:38444212	rs193922753	G|G	G	c.488G>T - Malignant Hyperthermia associated	
chr19:38446710	rs1801086	G|G	G	c.742G>A - Malignant Hyperthermia associated,c.742G>C - Malignant Hyperthermia associated	
chr19:38448673	rs193922762	C|C	C	c.982C>T - Malignant Hyperthermia associated	
chr19:38448712	rs121918592	G|G	G	c.1021G>A - Malignant Hyperthermia associated,c.1021G>C - Malignant Hyperthermia associated	
chr19:38451842	rs193922764	C|C	C	c.1201C>T - Malignant Hyperthermia associated	
chr19:38451850	rs118192116	C|C	C	c.1209C>G - Malignant Hyperthermia associated	
chr19:38455359	rs118192162	A|A	A	c.1565A>C - Malignant Hyperthermia associated	
chr19:38455463	rs111888148	G|G	G	c.1589G>A - Malignant Hyperthermia associated	

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Pharmacogenomics Report

John Doe (4376)

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chr19:38455471	rs193922768	C|C	C	c.1597C>T - Malignant Hyperthermia associated	
chr19:38455472	rs144336148	G|G	G	c.1598G>A - Uncertain function	
chr19:38455528	rs193922770	C|C	C	c.1654C>T - Malignant Hyperthermia associated	
chr19:38457545	rs118192172	C|C	C	c.1840C>T - Malignant Hyperthermia associated	
chr19:38457546	rs193922772	G|G	G	c.1841G>T - Malignant Hyperthermia associated	
chr19:38494564	rs118192175	C|C	C	c.6487C>T - Malignant Hyperthermia associated	
chr19:38494565	rs118192163	G|G	G	c.6488G>A - Malignant Hyperthermia associated	
chr19:38494579	rs118192176	G|G	G	c.6502G>A - Malignant Hyperthermia associated	
chr19:38496283	rs118192177	C|C	C	c.6617C>G - Malignant Hyperthermia associated,c.6617C>T - Malignant Hyperthermia associated	
chr19:38499223	rs112563513	G|G	G	c.7007G>A - Malignant Hyperthermia associated	
chr19:38499644	rs121918596	TGGA|TGGA	TGGA	c.7042_7044delGAG - Malignant Hyperthermia associated	

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Pharmacogenomics Report

John Doe (4376)

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chr19:38499655	rs193922802	G|G	G	c.7048G>A - Malignant Hyperthermia associated	
chr19:38499670	rs193922803	C|C	C	c.7063C>T - Malignant Hyperthermia associated	
chr19:38499731	rs193922807	G|G	G	c.7124G>C - Malignant Hyperthermia associated	
chr19:38499975	rs193922809	G|G	G	c.7282G>A - Malignant Hyperthermia associated	
chr19:38499993	rs121918593	G|G	G	c.7300G>A - Malignant Hyperthermia associated	
chr19:38499997	rs28933396	G|G	G	c.7304G>A - Malignant Hyperthermia associated	
chr19:38500636	rs118192124	C|C	C	c.7354C>T - Malignant Hyperthermia associated	
chr19:38500642	rs193922816	C|C	C	c.7360C>T - Malignant Hyperthermia associated	
chr19:38500643	rs118192122	G|G	G	c.7361G>A - Malignant Hyperthermia associated	
chr19:38500654	rs28933397	C|C	C	c.7372C>T - Malignant Hyperthermia associated	
chr19:38500655	rs121918594	G|G	G	c.7373G>A - Malignant Hyperthermia associated	

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Pharmacogenomics Report

John Doe (4376)

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chr19:38500898	rs118192178	C|C	C	c.7522C>G - Malignant Hyperthermia associated,c.7522C>T - Malignant Hyperthermia associated	
chr19:38500899	rs193922818	G|G	G	c.7523G>A - Malignant Hyperthermia associated	
chr19:38512321	rs193922832	G|G	G	c.9310G>A - Malignant Hyperthermia associated	
chr19:38543832	rs193922843	G|G	G	c.11969G>T - Malignant Hyperthermia associated	
chr19:38580004	rs118192167	A|A	A	c.14387A>G - Malignant Hyperthermia associated	
chr19:38580094	rs121918595	C|C	C	c.14477C>T - Malignant Hyperthermia associated	
chr19:38580114	rs193922876	C|C	C	c.14497C>T - Malignant Hyperthermia associated	
chr19:38580370	rs193922878	C|C	C	c.14512C>G - Malignant Hyperthermia associated	
chr19:38580403	rs118192168	G|G	G	c.14545G>A - Malignant Hyperthermia associated	
chr19:38580440	rs63749869	G|G	G	c.14582G>A - Uncertain function	
chr19:38584989	rs118192170	T|T	T	c.14693T>C - Malignant Hyperthermia associated	

Pharmacogenomics Report

John Doe (4376)

SLCO1B1 allele match data

Genotype matched: *37/*37
Phasing status: Unphased

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chr12:21172734	rs139257324	C|C	C	*33 - Uncertain function	
chr12:21172776	rs373327528	G|G	G	*23 - No function	
chr12:21172782	rs56101265	T|T	T	*2 - Uncertain function,*12 - Uncertain function	
chr12:21174595	rs56061388	T|T	T	*3 - Uncertain function,*13 - Uncertain function	
chr12:21176804	rs2306283	G/G	A	*14 - Increased function,*15 - No function,*20 - Increased function,*24 - Uncertain function,*25 - Uncertain function,*27 - Uncertain function,*28 - Uncertain function,*29 - Uncertain function,*30 - Uncertain function,*31 - No function,*32 - Uncertain function,*33 - Uncertain function,*37 - Normal function,*39 - Uncertain function,*42 - Uncertain function,*43 - Unknown function,*44 - Unknown function,*46 - No function,*47 - No function	

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Pharmacogenomics Report

John Doe (4376)

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chr12:21176868	rs2306282	A|A	A	*16 - Uncertain function	
chr12:21176871		G|G	G	*38 - Uncertain function	
chr12:21176879	rs11045819	C|C	C	*4 - Uncertain function,*14 - Increased function,*25 - Uncertain function,*32 - Uncertain function,*43 - Unknown function	
chr12:21176883	rs72559745	A|A	A	*3 - Uncertain function,*13 - Uncertain function	
chr12:21176898	rs77271279	G|G	G	*41 - Uncertain function	
chr12:21178612	rs141467543	A|A	A	*42 - Uncertain function	
chr12:21178615	rs4149056	T|T	T	*5 - No function,*15 - No function,*40 - Uncertain function,*46 - No function,*47 - No function	
chr12:21178957	rs79135870	A|A	A	*30 - Uncertain function	
chr12:21196951	rs11045852	A|A	A	*24 - Uncertain function,*25 - Uncertain function,*28 - Uncertain function,*32 - Uncertain function,*33 - Uncertain function,*43 - Unknown function,*44 - Unknown function	
chr12:21196975	rs183501729	C|C	C	*39 - Uncertain function	

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Pharmacogenomics Report

John Doe (4376)

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chr12:21196976	rs11045853	G|G	G	*25 - Uncertain function,*28 - Uncertain function,*33 - Uncertain function	
chr12:21200544	rs72559747	C|C	C	*47 - No function	
chr12:21200595	rs55901008	T|T	T	*6 - Uncertain function	
chr12:21202553	rs1228465562	T|T	T	*36 - Uncertain function	
chr12:21202555	rs59113707	C|C	C	*27 - Uncertain function	
chr12:21202649	rs56387224	A|A	A	*7 - Uncertain function	
chr12:21202664	rs142965323	G|G	G	*26 - Uncertain function	
chr12:21205921	rs72559748	A|A	A	*8 - Uncertain function	
chr12:21205999	rs59502379	G|G	G	*9 - No function,*31 - No function	
chr12:21206031	rs74064213	A|A	A	*43 - Unknown function,*44 - Unknown function	
chr12:21222355	rs71581941	C|C	C	*45 - Unknown function,*46 - No function	
chr12:21239042	rs34671512	A|A	A	*19 - Uncertain function,*20 - Increased function,*40 - Uncertain function	
chr12:21239077	rs56199088	A|A	A	*10 - Uncertain function,*12 - Uncertain function	
chr12:21239113	rs55737008	A|A	A	*11 - Uncertain function,*13 - Uncertain function	
chr12:21239145	rs200995543	C|C	C	*34 - Uncertain function	
chr12:21239158	rs140790673	C|C	C	*29 - Uncertain function	

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Pharmacogenomics Report

John Doe (4376)

TPMT allele match data

Genotype matched: *1/*1
Phasing status: Phased

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chr6:18130687	rs1142345	T|T	T	*3A - No function,*3C - No function,*41 - No function	
chr6:18130694	rs150900439	T|T	T	*20 - Uncertain function	
chr6:18130725	rs72552736	A|A	A	*7 - Uncertain function	
chr6:18130729	rs139392616	C|C	C	*40 - Uncertain function	
chr6:18130758	rs398122996	A|A	A	*37 - Uncertain function	
chr6:18130762	rs56161402	C|C	C	*8 - Uncertain function	
chr6:18130772	rs377085266	A|A	A	*25 - Uncertain function	
chr6:18130781	rs1800584	C|C	C	*4 - No function	
chr6:18132136	rs72556347	A|A	A	*26 - Uncertain function	
chr6:18132147	rs79901429	A|A	A	*31 - Uncertain function	
chr6:18132163		C|C	C	*36 - Unknown function	
chr6:18133845	rs75543815	T|T	T	*6 - Uncertain function	
chr6:18133847	rs6921269	C|C	C	*24 - Uncertain function	
chr6:18133870	rs772832951	A|A	A	*38 - Unknown function	
chr6:18133884	rs74423290	G|G	G	*23 - No function	
chr6:18133887	rs201695576	T|T	T	*44 - Unassigned function	
chr6:18133890	rs9333570	C|C	C	*15 - No function	

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Pharmacogenomics Report

John Doe (4376)

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chr6:18138969	rs144041067	C|C	C	*16 - Uncertain function,*22 - Uncertain function	
chr6:18138970	rs112339338	G|G	G	*33 - Uncertain function	
chr6:18138997	rs1800460	C|C	C	*3A - No function,*3B - No function	
chr6:18139027	rs72552737	C|C	C	*10 - Uncertain function	
chr6:18139689	rs72552738	C|C	C	*11 - No function	
chr6:18139710	rs200220210	G|G	G	*12 - Uncertain function	
chr6:18143597		T|T	T	*19 - Uncertain function	
chr6:18143606	rs151149760	T|T	T	*9 - Uncertain function	
chr6:18143613		C|C	C	*28 - Uncertain function	
chr6:18143622	rs115106679	C|C	C	*32 - Uncertain function	
chr6:18143643		A|A	A	*27 - Uncertain function	
chr6:18143700	rs753545734	C|C	C	*43 - Unassigned function	
chr6:18143718	rs111901354	G|G	G	*34 - Uncertain function	
chr6:18143724	rs1800462	C|C	C	*2 - No function	
chr6:18143728	rs1256618794	C|C	C	*43 - Unassigned function	
chr6:18147838	rs281874771	G|G	G	*39 - Uncertain function	
chr6:18147845	rs777686348	C|C	C	*18 - Uncertain function	
chr6:18147851	rs200591577	G|G	G	*21 - Uncertain function	
chr6:18147856		A|A	A	*35 - Unknown function	

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Pharmacogenomics Report

John Doe (4376)

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chr6:18147910	rs72552740	A|A	A	*5 - Uncertain function	
chr6:18149004		G|G	G	*17 - Uncertain function	
chr6:18149022	rs750424422	C|C	C	*30 - Unknown function	
chr6:18149032	rs759836180	C|C	C	*42 - Unassigned function	
chr6:18149045	rs72552742	T|T	T	*13 - Uncertain function	
chr6:18149126	rs267607275	A|A	A	*29 - No function	
chr6:18149127	rs9333569	T|T	T	*14 - No function	

UGT1A1 allele match data

Genotype matched: *1/*6
Phasing status: Unphased

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chr2:233759924	rs887829	C|C	C	*80 - Unknown function,*80+*28 - Decreased function,*80+*37 - Decreased function	
chr2:233760233	rs3064744	CAT|CAT	CAT	*28 - Decreased function,*36 - Increased function,*37 - Decreased function,*80+*28 - Decreased function,*80+*37 - Decreased function	
chr2:233760498	rs4148323	G/A	G	*6 - Decreased function	
chr2:233760973	rs35350960	C|C	C	*27 - Decreased function	

VKORC1 allele match data

Pharmacogenomics Report

John Doe (4376)

Genotype matched: rs9923231 variant (T)/rs9923231 variant (T)
Phasing status: Unphased

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chr16:31096368	rs9923231	T/T	C	rs9923231 variant (T) - Unassigned function	

Disclaimer and Other Information

novoClinic Disclaimer

The information provided in this report is intended for research and informational purposes only. While every effort has been made to ensure the accuracy of the data and interpretations derived from the CPIC database and PharmGKB-DPWG, the results generated by novoClinic should not be considered as a substitute for professional medical advice, diagnosis, or treatment.

It is important to note that pharmacogenetic analysis is a rapidly evolving field, and interpretations of genetic variants may change over time as new evidence emerges. Additionally, individual responses to medications can vary due to factors beyond genetic variations, including environmental influences and other health conditions.

The recommendations provided in this report are based on the best available evidence at the time of analysis and may not encompass all possible scenarios or considerations relevant to a patient's medical care. Healthcare providers should exercise their clinical judgment and consider the unique circumstances of each patient when making treatment decisions.

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CPIC Allele Function, Phenotype and Recommendation

The entirety of the content originates from the CPIC database, accessible at https://cpicpgx.org/.

DPWG Allele Function, Phenotype and Recommendation

PharmGKB provides annotations for pharmacogenomics (PGx) based drug dosing guidelines issued by the Royal Dutch Association for the Advancement of Pharmacy - Pharmacogenetics Working Group (DPWG). PharmGKB meticulously curates allele function assignments and phenotype mappings derived from DPWG data to provide genotype-specific DPWG guideline recommendations. Whenever feasible, PharmGKB aligns DPWG terminology with CPIC terms, as outlined on PharmGKB.

Genes

Table below shows the list of genes tested by novoClinic.

ABCG2	CYP2C19	CYP4F2	NUDT15	UGT1A1
CACNA1S	CYP2C9	DPYD	RYR1	VKORC1
CFTR	CYP3A4	F5	SLCO1B1	
CYP2B6	CYP3A5	G6PD	TPMT	

Drugs

Table below shows the list of drugs tested by novoClinic.

abacavir	dexlansoprazole	irinotecan	peginterferon alfa-2a	streptomycin
acenocoumarol	doxepin	isoflurane	peginterferon alfa-2b	succinylcholine
allopurinol	efavirenz	ivacaftor	pegloticase	tacrolimus
amikacin	eliglustat	kanamycin	phenprocoumon	tafenoquine
amitriptyline	enflurane	lamotrigine	phenytoin	tamoxifen
aripiprazole	escitalopram	lansoprazole	pimozide	tegafur
atazanavir	flecainide	lornoxicam	piroxicam	tenoxicam
atomoxetine	flucloxacillin	lovastatin	pitavastatin	thioguanine
atorvastatin	flucytosine	meloxicam	plazomicin	tobramycin
azathioprine	fluorouracil	mercaptopurine	pravastatin	toluidine blue
brexpiprazole	flurbiprofen	methoxyflurane	primaquine	tramadol
capecitabine	fluvastatin	metoprolol	propafenone	trimipramine
carbamazepine	fluvoxamine	methylene blue	quetiapine	tropisetron
celecoxib	fosphenytoin	nitrofurantoin	rasburicase	venlafaxine
citalopram	gentamicin	nortriptyline	ribavirin	voriconazole
clomipramine	haloperidol	omeprazole	risperidone	vortioxetine
clopidogrel	halothane	ondansetron	rosuvastatin	warfarin
codeine	hormonal contraceptives for systemic use	oxcarbazepine	sertraline	zuclopenthixol
dapsone	hydrocodone	pantoprazole	sevoflurane	
desflurane	ibuprofen	paromomycin	simvastatin	
desipramine	imipramine	paroxetine	siponimod	